

# GENELIST GENE PANELS DG 3.4.0 (4874 genes)

Releasedate: 19-04-2022

<i>Gene</i>	<i>Genepanels</i>	<i>TWIST covered &gt;10x</i>	<i>TWIST covered &gt;20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
A2M	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
A2ML1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
A4GALT	MENDELIOME	100,0%	100,0%	NOR polyagglutination syndrome, 111400
AAAS	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	NOR polyagglutination syndrome, 111400
AAGAB	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287 ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 Trichothiodystrophy 8, nonphotosensitive, 619691
AARS2	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096

AASS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperlysinemia, 238700
ABAT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	GABA-transaminase deficiency, 613163
ABCA1	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA12	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,5%	96,5%	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200
ABCA5	MENDELIOME	100,0%	100,0%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB10	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ABCB11	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347

ABCB6	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Microphthalmia, isolated, with coloboma 7, 614497 Dyschromatosis universalis hereditaria 3, 615402 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCB7	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME	99,7%	99,5%	Anemia, sideroblastic, with ataxia, 301310
ABCC1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 77, 618915
ABCC2	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dubin-Johnson syndrome, 237500
ABCC6	VISION DISORDERS SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 ?Atrial fibrillation, familial, 12, 614050 Intellectual disability and myopathy syndrome, 619719

ABCD1	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME	100,0%	100,0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ABCD3	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sitosterolemia 2, 618666
ABCG8	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sitosterolemia 1, 210250
ABHD12	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674

ABHD16A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chanarin-Dorfman syndrome, 275630
ABL1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACACA	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
ACAD8	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	2-methylbutyrylglycinuria, 610006

ACADVL	HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	VLCAD deficiency, 201475
ACAN	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	98,7%	98,6%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACAT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ACBD5	VISION DISORDERS HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Retinal dystrophy with leukodystrophy, 618863
ACD	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACE	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal tubular dysgenesis, 267430

ACER3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACKR3	MENDELIOME	100,0%	100,0%	?Oculomotor-abducens synkinesis, 619215
ACO2	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	Amelogenesis imperfecta, type IJ, 617297
ACP5	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 63, 300387
ACSL6	MENDELIOME	97,1%	97,1%	Myelodysplastic syndrome, Myelogenous leukemia, acute,

ACTA1	FETAL AKINESIA (AKI) MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ACTA2	SKIN DISORDERS LIVER DISORDERS MENDELIOME	100,0%	100,0%	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042
ACTB	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTC1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424
ACTG1	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACTG2	LIVER DISORDERS MENDELIOME	100,0%	100,0%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 Visceral myopathy 1, 155310
ACTL6A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

ACTL6B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTL9	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 53, 619258
ACTN1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	HEART MENDELIOME	100,0%	100,0%	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACTN4	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	IRON DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	MENDELIOME	100,0%	100,0%	Pancreatic cancer, somatic,
ACVR2B	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME	100,0%	100,0%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 2, 600376

ACY1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Aminoacylase 1 deficiency, 609924
ADA	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAD2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADAM10	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Reticulate acropigmentation of Kitamura, 615537
ADAM17	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 61, 617933
ADAM9	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 9, 612775
ADAMTS1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID

ADAMTS10	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS17	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS19	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADAMTS2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	98,1%	98,1%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTS9	CILIOPATHIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADAMTSL2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,7%	Geleophysic dysplasia 1, 231050

ADAMTSL4	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	MOVEMENT DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADARB1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	95,1%	95,1%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADCK2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ADCK5	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADCY1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	99,3%	98,9%	?Deafness, autosomal recessive 44, 610154
ADCY10	MALE INFERTILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADCY3	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADCY5	MOVEMENT DISORDERS HEART METABOLIC DISORDERS MENDELIOME	100,0%	99,9%	Dyskinesia with orofacial involvement, autosomal dominant, 606703 Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 Dyskinesia with orofacial involvement, autosomal recessive, 619647

ADCY6	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lethal congenital contracture syndrome 8, 616287
ADD3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	MENDELIOME	99,3%	98,7%	Vibratory urticaria, 125630
ADGRG1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
ADH5	MENDELIOME	100,0%	100,0%	AMED syndrome, digenic, 619151
ADIPOQ	MENDELIOME	100,0%	100,0%	Adiponectin deficiency, 612556
ADIPOR1	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ADK	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	84,5%	84,5%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	INTELLECTUAL DISABILITY MENDELIOME	95,4%	95,4%	Helsmoortel-van der Aa syndrome, 615873

ADPRS	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRB2	MENDELIOME	100,0%	100,0%	Beta-2-adrenoreceptor agonist, reduced response to,
ADSL	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adenylosuccinase deficiency, 103050
ADSS1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, distal, 5, 617030
AEBP1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	KINSHIP syndrome, 619297
AFF4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	CHOPS syndrome, 616368
AFG3L2	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AFP	MENDELIOME	100,0%	100,0%	Alpha-fetoprotein deficiency, 615969

AGA	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Aspartylglucosaminuria, 208400
AGAP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
AGBL1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 75, 617023
AGK	VISION DISORDERS HEART METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	91,2%	91,2%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGMO	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
AGO1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

AGO2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Lessel-Kreienkamp syndrome, 619149
AGPAT2	SKIN DISORDERS HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal tubular dysgenesis, 267430
AGTPBP1	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal tubular dysgenesis, 267430
AGXT	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperoxaluria, primary, type 1, 259900

AHCY	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Xia-Gibbs syndrome, 615829
AHI1	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 3, 608629
AHNAK2	NEUROPATHIES MENDELIOME	97,7%	97,6%	No OMIM Disease ID
AHR	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Retinitis pigmentosa 85, 618345
AHSG	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Alopecia-intellectual disability syndrome 1, 203650
AICDA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	HEARING IMPAIRMENT NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
AIMP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 3, 260600

AIMP2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 17, 618006
AIP	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AIPL1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Reticular dysgenesis, 267500
AK7	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 27, 617965
AKAP9	HEART MENDELIOME	100,0%	100,0%	?Long QT syndrome 11, 611820
AKR1C1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
AKR1C2	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	46XY sex reversal 8, 614279
AKR1D1	LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bile acid synthesis defect, congenital, 2, 235555

AKT1	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Colorectal cancer, somatic, 114500 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000
AKT2	NEWBORN BLOOD SPOT SCREENING MENDELIOME	100,0%	100,0%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Porphyria, acute hepatic, 612740
ALAS2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Anemia, sideroblastic, 1, 300751 Protoporphyrina, erythropoietic, X-linked, 300752
ALB	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Analbuminemia, 616000
ALDH18A1	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraparesis 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraparesis 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603
ALDH1A2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID

ALDH1A3	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, isolated 8, 615113
ALDH1B1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ALDH2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Alcohol sensitivity, acute, 610251
ALDH3A2	MOVEMENT DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	93,2%	93,2%	Sjogren-Larsson syndrome, 270200
ALDH4A1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperprolinemia, type II, 239510
ALDH5A1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycogen storage disease XII, 611881

ALDOB	SKIN DISORDERS LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fructose intolerance, hereditary, 229600
ALG1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type I <sub>k</sub> , 608540
ALG10	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ALG11	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	96,8%	96,8%	Congenital disorder of glycosylation, type I <sub>p</sub> , 613661
ALG12	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type I <sub>g</sub> , 607143
ALG13	EPILEPSY PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Developmental and epileptic encephalopathy 36, 300884
ALG14	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227

ALG2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	FETAL AKINESIA (AKI) EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,6%	96,6%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type II, 608776
ALK	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
ALKBH1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ALKBH8	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 71, 618504

ALMS1	VISION DISORDERS CILIOPATHIES HEARING IMPAIRMENT HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alstrom syndrome, 203800
ALOX12B	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPI	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ALPK1	MENDELIOME	100,0%	100,0%	ROSAH syndrome, 614979
ALPK3	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
ALS2	ALS MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100

ALX1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Frontonasal dysplasia 3, 613456
ALX3	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Frontonasal dysplasia 1, 136760
ALX4	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMACR	EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBН	CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IF, 616270
AMELX	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	100,0%	100,0%	Amelogenesis imperfecta, type 1E, 301200
AMER1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Osteopathia striata with cranial sclerosis, 300373

AMH	DISORDERS OF SEX DEVELOPMENT MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	DISORDERS OF SEX DEVELOPMENT MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMPD3	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
AMT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycine encephalopathy, 605899
AMTN	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	?Amelogenesis imperfecta, type IIIB, 617607

ANAPC1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Rothmund-Thomson syndrome, type 1, 618625
ANAPC7	MENDELIOME	100,0%	100,0%	Ferguson-Bonni neurodevelopmental syndrome, 619699
ANG	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 9, 611895
ANGPT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Angioedema, hereditary, 5, 619361
ANGPT2	MENDELIOME	100,0%	100,0%	Lymphatic malformation 10, 619369
ANGPTL3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	MENDELIOME	100,0%	100,0%	Plasma triglyceride level QTL, low, 615881
ANK1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spherocytosis, type 1, 182900
ANK2	HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 37, 615493
ANKFY1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ANKH	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 16, primary, autosomal recessive, 616681

ANKRD1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ANKRD11	CRANIOFACIAL ANOMALIES SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	KBG syndrome, 148050
ANKRD17	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Chopra-Amiel-Gordon syndrome, 619504
ANKRD26	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME HEREDITARY CANCER	97,2%	97,2%	Thrombocytopenia 2, 188000
ANKS1B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ANKS6	CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Nephronophthisis 16, 615382
ANLN	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	100,0%	100,0%	Focal segmental glomerulosclerosis 8, 616032
ANO10	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 10, 613728

AN03	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Dystonia 24, 615034
AN05	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoji muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
AN06	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Scott syndrome, 262890
AN081	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	GAPO syndrome, 230740
ANTXR2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyaline fibromatosis syndrome, 228600
ANXA11	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 23, 617839 Inclusion body myopathy and brain white matter abnormalities, 619733
AOPEP	MENDELIOME	100,0%	100,0%	Dystonia 31, 619565
AP1B1	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150

AP1G1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Usmani-Riazuddin syndrome, autosomal recessive, 619548 Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	MEDNIK syndrome, 609313
AP1S2	INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,9%	Pettigrew syndrome, 304340
AP1S3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	90,5%	90,5%	No OMIM Disease ID
AP2M1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,7%	Developmental and epileptic encephalopathy 48, 617276

AP3D1	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	87,9%	87,9%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 48, autosomal recessive, 613647
APC	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Desmoid disease, hereditary, 135290 Adenoma, periamppullary, somatic, 175100 Hepatoblastoma, somatic, 114550 Gastric cancer, somatic, 613659 Gastric adenocarcinoma and proximal polyposis of the stomach, 619182 Gardner syndrome, 175100 Adenomatous polyposis coli, 175100
APC2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 10, 618677 Intellectual developmental disorder, autosomal recessive 74, 617169

APCDD1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Hypotrichosis 1, 605389
APOA1	MENDELIOME	100,0%	100,0%	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 618463
APOA2	MENDELIOME	100,0%	100,0%	Apolipoprotein A-II deficiency,
APOA5	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Hyperchylomicronemia, late-onset, 144650
APOB	MENDELIOME	100,0%	100,0%	Hypercholesterolemia, familial, 2, 144010 Hypobetalipoproteinemia, 615558
APOC2	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperlipoproteinemia, type Ib, 207750
APOC3	MENDELIOME	100,0%	100,0%	Apolipoprotein C-III deficiency, 614028
APOE	NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alzheimer disease 2, 104310 Sea-blue histiocyte disease, 269600 Lipoprotein glomerulopathy, 611771 Hyperlipoproteinemia, type III, 617347
APOL1	PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
APOO	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
APP	MENDELIOME	100,0%	100,0%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
APRT	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adenine phosphoribosyltransferase deficiency, 614723

APTX	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diabetes insipidus, nephrogenic, 2, 125800
AQPS	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Palmoplantar keratoderma, Bothnian type, 600231
AR	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME	100,0%	100,0%	Androgen insensitivity, partial, with or without breast cancer, 312300 Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Hypospadias 1, X-linked, 300633
ARCN1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	97,0%	96,6%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Periventricular nodular heterotopia 8, 618185
ARF3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARFGEF1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARFGEF2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Periventricular heterotopia with microcephaly, 608097

ARG1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	92,9%	92,9%	Argininemia, 207800
ARHGAP24	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARHGAP26	MENDELIOME	100,0%	100,0%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP29	CRANIOFACIAL ANOMALIES MENDELIOME SCHISIS	100,0%	100,0%	No OMIM Disease ID
ARHGAP31	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Adams-Oliver syndrome 1, 100300
ARHGAP35	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARHGDIA	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 8, 615244
ARHGEF1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency 62, 618459
ARHGEF10	NEUROPATHIES MENDELIOME	100,0%	100,0%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 78, 617433
ARHGEF2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523

ARHGEF28	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARHGEF6	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARHGEF9	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	97,2%	97,2%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 2, 614607
ARID1B	SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	98,6%	98,3%	Coffin-Siris syndrome 1, 135900
ARID2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 6, 617808
ARIH1	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARL13B	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 8, 612291
ARL2	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa with or without situs inversus, 615434

ARL3	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARL6IP1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 38, 618433
ARMC5	DISORDERS OF SEX DEVELOPMENT MENDELIOME HEREDITARY CANCER	100,0%	100,0%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 30, 617622
ARNT2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Webb-Dattani syndrome, 615926
ARPC1B	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARPC4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ARR3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Myopia 26, X-linked, female-limited, 301010

ARSA	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Metachromatic leukodystrophy, 250100
ARSB	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Usher syndrome, type IV, 618144
ARSK	MENDELIOME	100,0%	100,0%	Mucopolysaccharidosis, type X, 619698
ARSL	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 38, 617020
ARX	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99,0%	96,8%	Proud syndrome, 300004 Hydranencephaly with abnormal genitalia, 300215 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Lissencephaly, X-linked 2, 300215 Intellectual developmental disorder, X-linked 29, 300419
ASAHI	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000

ASB10	VISION DISORDERS MENDELIOME	100,0%	100,0%	Glaucoma 1, open angle, F, 603383
ASCC1	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	87,1%	87,1%	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ASH1L	INTELLECTUAL DISABILITY MENDELIOME	98,7%	98,7%	Intellectual developmental disorder, autosomal dominant 52, 617796
ASIP	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ASL	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Argininosuccinic aciduria, 207900
ASNS	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Asparagine synthetase deficiency, 615574
ASPA	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Canavan disease, 271900
ASPH	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Traboulsi syndrome, 601552
ASPM	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 5, primary, autosomal recessive, 608716

ASPRV1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Ichthyosis, lamellar, autosomal dominant, 146750
ASPSCR1	MENDELIOME	100,0%	100,0%	Alveolar soft-part sarcoma, 606243
ASRGL1	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ASS1	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Citrullinemia, 215700
ASTL	MENDELIOME	100,0%	100,0%	?Oocyte maturation defect 11, 619643
ASXL1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS HEREDITARY CANCER	99,9%	99,9%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ASXL2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Shashi-Pena syndrome, 617190
ASXL3	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Bainbridge-Ropers syndrome, 615485
ATAD1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperekplexia 4, 618011

ATAD3A	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATCAY	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ataxia, cerebellar, Cayman type, 601238
ATF3	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATF6	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Achromatopsia 7, 616517
ATG4A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	99,9%	No OMIM Disease ID
ATG5	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATG7	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	MOVEMENT DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spastic paraparesis 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	Neuropathy, hereditary sensory, type IF, 615632

ATM	MOVEMENT DISORDERS BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATN1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATOH7	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11A	MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 84, 619810
ATP11C	MENDELIOME	100,0%	99,9%	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraparesis 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A1	EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036

ATP1A2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 98, 619605 Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PARKINSON	100,0%	100,0%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
ATP2A1	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Brody myopathy, 601003
ATP2A2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2B2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 82, 619804
ATP2B3	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Hailey-Hailey disease, 169600
ATP4A	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATP5F1A	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1B	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5F1C	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

ATP5F1D	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MC1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MC2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MC3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Dystonia, early-onset, and/or spastic paraplegia, 619681
ATP5ME	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MF	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MG	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MGL	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5MD	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5PD	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5PF	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP5PO	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATP6AP1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Immunodeficiency 47, 300972

ATP6AP2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATP6V0A2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V0C	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATP6V1A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1B1	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1E1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIC, 617402

ATP7A	SKIN DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
ATP7B	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Wilson disease, 277900
ATP8A2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300
ATP9A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATPAF1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
ATPAF2	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273

ATR	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Intellectual disability-hypotonic facies syndrome, X-linked, 309580
ATXN1	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 1, 164400
ATXN10	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 10, 603516
ATXN2	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 2, 183090
ATXN2L	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ATXN3	MENDELIOME	95,8%	95,8%	Machado-Joseph disease, 109150
ATXN7	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 7, 164500
ATXN8OS	MENDELIOME	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	3-methylglutaconic aciduria, type I, 250950
AURKC	MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spermatogenic failure 5, 243060

AUTS2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVIL	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephrotic syndrome, type 21, 618594
AVP	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Diabetes insipidus, nephrogenic, 1, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	MENDELIOME	100,0%	100,0%	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864
AXIN2	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
AXL	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	No OMIM Disease ID
B2M	NEUROPATHIES PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B3GALNT1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
B3GALNT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	92,5%	92,5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181

B3GALT6	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	99,8%	98,8%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GAT3	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,2%	94,9%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Peters-plus syndrome, 261540
B4GALNT1	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALNT2	MENDELIOME	100,0%	100,0%	Sd(a) polyagglutination syndrome, 615018
B4GALT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type II <sup>d</sup> , 607091
B4GALT7	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070

B4GAT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	CILIOPATHIES DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,6%	94,1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bile acid conjugation defect 1, 619232
BACH2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 60 and autoimmunity, 618394
BAG3	HEART NEUROPATHIES MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BAG5	MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 2F, 619747
BANF1	SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nestor-Guillermo progeria syndrome, 614008

BAP1	BREAST CANCER SKIN DISORDERS MELANOOM INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Tumor predisposition syndrome, 614327 Kury-Isidor syndrome, 619762
BARD1	BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
BAX	MENDELIOME	100,0%	100,0%	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BAZ2B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BBIP1	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Bardet-Biedl syndrome 18, 615995
BBS1	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 1, 209900
BBS10	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 10, 615987
BBS12	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 12, 615989

BBS2	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 4, 615982
BBS5	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 5, 615983
BBS7	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 7, 615984
BBS9	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	95,8%	95,8%	Bardet-Biedl syndrome 9, 615986
BCAP31	MOVEMENT DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Hengel-Maroffian-Schols syndrome, 619641

BCAT1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BCAT2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	?Hypervalinemia or hyperleucine-isoleucinemia, 618850
BCHE	MENDELIOME	100,0%	100,0%	Butyrylcholinesterase deficiency, 617936
BCKDHA	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Maple syrup urine disease, type Ia, 248600
BCKDHB	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Maple syrup urine disease, type Ib, 248600
BCKDK	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
BCL10	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Dias-Logan syndrome, 617101
BCL11B	MOVEMENT DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 Immunodeficiency 49, 617237

BCL2	MENDELIOME	100,0%	100,0%	Leukemia/lymphoma, B-cell, 2,
BCL7A	MENDELIOME	100,0%	100,0%	B-cell non-Hodgkin lymphoma, high-grade,
BCO1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Microphthalmia, syndromic 2, 300166
BCORL1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Shukla-Vernon syndrome, 301029
BCS1L	SKIN DISORDERS HEARING IMPAIRMENT LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BDP1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal recessive 112, 618257
BEAN1	MENDELIOME	92,2%	92,2%	Spinocerebellar ataxia 31, 117210
BEST1	VISION DISORDERS MENDELIOME	100,0%	99,9%	Macular dystrophy, vitelliform, 2, 153700 ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Bestrophinopathy, autosomal recessive, 611809
BFSP1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 33, multiple types, 611391

BFSP2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 12, multiple types, 611597
BGN	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BICC1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BICD2	FETAL AKINESIA (AKI) NEUROPATHIES MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BICRA	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 12, 619325
BIN1	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Centronuclear myopathy 2, 255200
BLK	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Maturity-onset diabetes of the young, type 11, 613375
BLM	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Bloom syndrome, 210900

BLNK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Agammaglobulinemia 4, 613502
BLOC1S1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BLOC1S3	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Hermansky-Pudlak syndrome 9, 614171
BLVRA	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperbiliverdinemia, 614156
BMP1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XIII, 614856
BMP15	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Premature ovarian failure 4, 300510 Ovarian dysgenesis 2, 300510

BMP2	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMP4	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BMP6	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BMP7	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BMPer	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Diaphanospondylodysostosis, 608022
BMPR1A	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acromesomelic dysplasia 3, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849
BMPR2	HEART MENDELIOME	99,9%	99,9%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BMS1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Aplasia cutis congenita, nonsyndromic, 107600
BNC1	MENDELIOME	100,0%	99,9%	?Premature ovarian failure 16, 618723

BNC2	MENDELIOME	100,0%	100,0%	Lower urinary tract obstruction, congenital, 618612
BOLA1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
BOLA2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
BOLA3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Erythrocytosis, familial, 8, 222800
IMPAD1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BPTF	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980

BRAT1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	HEREDITARY BONE MARROW FAILURE BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group S, 617883
BRCA2	HEREDITARY BONE MARROW FAILURE BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRDT	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 21, 617644
BRF1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellofaciodental syndrome, 616202
BRIP1	HEREDITARY BONE MARROW FAILURE BREAST CANCER SKIN DISORDERS HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group J, 609054

BRPF1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BRWD3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 93, 300659
BSCL2	MOVEMENT DISORDERS SKIN DISORDERS HEART NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	83,1%	83,1%	Biotinidase deficiency, 253260
BTG4	MENDELIOME	100,0%	100,0%	Oocyte maturation defect 8, 619009
BTK	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200

BTRC	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
BUB1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
BVES	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C11orf80	MENDELIOME	91,9%	91,9%	Hydatidiform mole, recurrent, 4, 618432
C12orf4	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 66, 618221
C12orf57	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Temptamy syndrome, 218340
C14orf39	MENDELIOME	100,0%	100,0%	Spermatogenic failure 52, 619202 ?Premature ovarian failure 18, 619203
C19orf12	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1GALT1C1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Tn polyagglutination syndrome, somatic, 300622
C1QA	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C1q deficiency, 613652

C1QB	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C1q deficiency, 613652
C1QBP	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C1q deficiency, 613652
C1QTNF5	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	99,1%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,2%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	C2 deficiency, 217000
C2CD3	CILIOPATHIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	95,9%	95,9%	Orofaciodigital syndrome XIV, 615948
C2CD6	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 68, 619805
C2orf69	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 53, 619423

C3	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C3 deficiency, 613779
C4A	MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	C4a deficiency, 614380
C4B	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C4B deficiency, 614379
C5	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C5 deficiency, 609536
C6	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	C6 deficiency, 612446 Combined C6/C7 deficiency,
C7	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	C7 deficiency, 610102
C8A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C8 deficiency, type I, 613790
C8B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C8 deficiency, type II, 613789
C8G	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
C9	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	C9 deficiency, 613825
C9orf72	MENDELIOME	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperchlorhidrosis, isolated, 143860

CA2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CA5A	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	87,7%	87,7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABIN1	HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CABP2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 93, 614899
CABP4	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	MOVEMENT DISORDERS EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Developmental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497

CACNA1C	HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	HEARING IMPAIRMENT HEART INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 69, 618285
CACNA1F	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA1G	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hyperaldosteronism, familial, type IV, 617027
CACNA1I	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CACNA1S	FETAL AKINESIA (AKI) MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Hypokalemic periodic paralysis, type 1, 170400
CACNA2D1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CACNA2D2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar atrophy with seizures and variable developmental delay, 618501

CACNA2D4	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinal cone dystrophy 4, 610478
CACNB2	HEART MENDELIOME	100,0%	100,0%	Brugada syndrome 4, 611876
CACNB4	MOVEMENT DISORDERS EPILEPSY MENDELIOME	100,0%	100,0%	Episodic ataxia, type 5, 613855
CACNG2	MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder, autosomal dominant 10, 614256
CAD	HEREDITARY BONE MARROW FAILURE EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 50, 616457
CADM3	NEUROPATHIES MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519
CALCRL	MENDELIOME	100,0%	100,0%	?Lymphatic malformation 8, 618773
CALM1	HEART MENDELIOME	100,0%	100,0%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247
CALM2	HEART MENDELIOME	72,0%	72,0%	Long QT syndrome 15, 616249
CALM3	HEART MENDELIOME	100,0%	100,0%	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782

CALR	HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME	100,0%	100,0%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Intellectual developmental disorder, autosomal dominant 53, 617798 ?Intellectual developmental disorder, autosomal recessive 63, 618095
CAMK2B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 54, 617799
CAMK2G	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 59, 618522
CAMK4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CAMTA1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CANT1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
CAPN12	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CAPN15	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Oculogastrointestinal neurodevelopmental syndrome, 619318

CAPN3	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	97,9%	97,9%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	VISION DISORDERS MENDELIOME	100,0%	100,0%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD10	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency 89 and autoimmunity, 619632
CARD11	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD14	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD8	MENDELIOME	100,0%	100,0%	?Inflammatory bowel disease (Crohn disease) 30, 619079
CARD9	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 58, 618131
CARS1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, with or without nystagmus, 300422 Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422

CASP10	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP14	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	95,6%	95,6%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271
CASQ1	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	EPILEPSY HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CAST	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CASZ1	HEART MENDELIOME	99,8%	99,0%	No OMIM Disease ID
CAT	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acatalasemia, 614097
CATIP	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 54, 619379
CATSPER1	MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spermatogenic failure 7, 612997

CATSPER2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CAV1	SKIN DISORDERS HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAV3	HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myopathy, distal, Tateyama type, 614321 Creatine phosphokinase, elevated serum, 123320 Cardiomyopathy, familial hypertrophic, 192600 Rippling muscle disease 2, 606072 Long QT syndrome 9, 611818
CAVIN1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBLIF	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intrinsic factor deficiency, 261000

CBS	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CBWD1	RENAL DISORDERS MENDELIOME	99,4%	98,5%	No OMIM Disease ID
CBX2	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?46XY sex reversal 5, 613080
CBY1	CILIOPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CC2D1A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 3, 608443
CC2D2A	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	97,1%	97,1%	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCBE1	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 17, 614679

CCDC115	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIo, 616828
CCDC134	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Osteogenesis imperfecta, type XXII, 619795
CCDC141	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CCDC174	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CCDC22	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Ritscher-Schinzl syndrome 2, 300963
CCDC28B	CILIOPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CCDC32	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cardiofaciocorectal syndrome, 619123
CCDC39	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Trichohepatoneurodevelopmental syndrome, 618268

CCDC50	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 44, 607453
CCDC62	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 67, 619803
CCDC65	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	?Centronuclear myopathy 4, 614807
CCDC8	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	3-M syndrome 3, 614205
CCDC88A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	97,5%	97,5%	?PEHO syndrome-like, 617507
CCDC88C	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCL2	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CCM2	MENDELIOME	100,0%	100,0%	Cerebral cavernous malformations-2, 603284
CCN6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	85,4%	84,6%	Progressive pseudorheumatoid dysplasia, 208230
CCND2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNF	MENDELIOME	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141
CCNK	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147

CCNO	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 29, 615872
CCNQ	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	99,9%	99,8%	STAR syndrome, 300707
CCT2	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CCT5	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	SKIN DISORDERS HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 66, 616969
CD19	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 3, 613493
CD247	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	?Immunodeficiency 25, 610163
CD27	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Lymphoproliferative syndrome 2, 615122
CD28	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

CD2AP	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Platelet glycoprotein IV deficiency, 608404
CD3D	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 19, 615617
CD3E	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949
CD40	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency, X-linked, with hyper-IgM, 308230

CD46	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CD48	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CD55	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	95,9%	93,9%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	64,5%	64,5%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Lymphoproliferative syndrome 3, 618261
CD79A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Agammaglobulinemia 3, 613501
CD79B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Agammaglobulinemia 6, 612692
CD81	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 6, 613496
CD8A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	CD8 deficiency, familial, 608957
CD96	MENDELIOME	100,0%	100,0%	C syndrome, 211750

CDAN1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	HEARING IMPAIRMENT MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC40	MENDELIOME	100,0%	100,0%	?Pontocerebellar hypoplasia, type 15, 619302
CDC42	HEARING IMPAIRMENT HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CDC45	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Meier-Gorlin syndrome 7, 617063
CDC6	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Meier-Gorlin syndrome 5, 613805
CDC73	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001

CDCA7	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	BREAST CANCER MENDELIOME SCHISIS HEREDITARY CANCER	97,8%	97,8%	Ovarian cancer, somatic, 167000 Blepharocheilodontic syndrome 1, 119580 Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215 Endometrial carcinoma, somatic, 608089 Breast cancer, lobular, somatic, 114480
CDH11	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Teebi hypertelorism syndrome 2, 619736 Elsahy-Waters syndrome, 211380
CDH15	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 3, 612580
CDH2	VISION DISORDERS DISORDERS OF SEX DEVELOPMENT HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDH23	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386
CDH3	VISION DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDH4	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CDHR1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660

C15orf41	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Al Kaissi syndrome, 617694
CDK13	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 87, 618916
CDK4	SKIN DISORDERS MELANOMA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
CDK5	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 12, primary, autosomal recessive, 616080
CDK8	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	92,3%	92,2%	Developmental and epileptic encephalopathy 2, 300672
CDKN1A	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
CDKN1B	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Multiple endocrine neoplasia, type IV, 610755

CDKN1C	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	SKIN DISORDERS MELANOMA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
CDKN2B	PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
CDKN2C	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
CDON	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Holoprosencephaly 11, 614226
CDSN	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Meier-Gorlin syndrome 4, 613804
CEACAM16	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 4B, 614614 Deafness, autosomal recessive 113, 618410
CEBPA	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Specific granule deficiency, 245480

CEL	METABOLIC DISORDERS MENDELIOME	100,0%	99,9%	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	MENDELIOME	100,0%	100,0%	Abdominal obesity-metabolic syndrome 4, 618620
CELF2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 97, 619561
CELSR1	SKIN DISORDERS MENDELIOME	99,9%	99,9%	Lymphatic malformation 9, 619319
CENPE	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Stromme syndrome, 243605
CENPJ	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CENPT	MENDELIOME	100,0%	100,0%	?Short stature and microcephaly with genital anomalies, 618702
CEP104	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 25, 616781
CEP112	MENDELIOME	100,0%	100,0%	Spermatogenic failure 44, 619044

CEP120	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP135	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 15, 614845
CEP19	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Morbid obesity and spermatogenic failure, 615703
CEP250	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134

CEP41	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 15, 614464
CEP55	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	M multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Seckel syndrome 6, 614728
CEP78	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy and hearing loss, 617236
CEP83	VISION DISORDERS CILIOPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 18, 615862
CEP85L	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Lissencephaly 10, 618873
CEP89	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

CERKL	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 26, 608380
CERS1	MENDELIOME PRE CONCEPTION SCREENING	99,4%	98,1%	Epilepsy, progressive myoclonic, 8, 616230
CERS3	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 9, 615023
CERT1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 34, 616351
CES1	MENDELIOME	99,9%	99,8%	Drug metabolism, altered, CES1-related, 618057
CETP	MENDELIOME	100,0%	100,0%	Hyperalphalipoproteinemia, 143470
WDR66	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 33, 618152
C1orf194	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CFAP298	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	CILIOPATHIES MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 38, 618063
CFAP410	ALS VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271

C8orf37	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406
CFAP43	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Hydrocephalus, normal pressure, 1, 236690 Spermatogenic failure 19, 617592
CFAP44	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 20, 617593
CFAP45	MENDELIOME	100,0%	100,0%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP47	MENDELIOME	100,0%	100,0%	Spermatogenic failure, X-linked 3, 301059
CFAP52	MENDELIOME	100,0%	100,0%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP58	MALE INFERTILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Spermatogenic failure 49, 619144
CFAP65	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 40, 618664
CFAP69	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 24, 617959
CFAP70	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 41, 618670
MAATS1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 51, 619177

CFB	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	?Complement factor B deficiency, 615561
CFC1	CONGENITAL HEART DISEASE CILIOPATHIES HEART LIVER DISORDERS MENDELIOME	100,0%	100,0%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Complement factor D deficiency, 613912
CFH	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	RENAL DISORDERS MENDELIOME	93,0%	91,8%	No OMIM Disease ID
CFHR3	RENAL DISORDERS MENDELIOME	91,9%	91,1%	No OMIM Disease ID
CFHR5	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephropathy due to CFHR5 deficiency, 614809
CFI	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Complement factor I deficiency, 610984

CFL2	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Properdin deficiency, X-linked, 312060
CFTR	PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS MALE INFERTILITY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHAMP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 40, 616579
CHAT	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	ALS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	100,0%	100,0%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON	100,0%	100,0%	Parkinson disease 22, autosomal dominant, 616710
CHD1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Pilarowski-Bjornsson syndrome, 617682
CHD2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 94, 615369
CHD3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Snijders Blok-Campeau syndrome, 618205

CHD4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CHD7	VISION DISORDERS CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT HEART HYPOGONADOTROPIC HYPOGONADISM PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CHEK2	BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Osteosarcoma, somatic, 259500 Li-Fraumeni syndrome 2, 609265
CHIT1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CHKB	SKIN DISORDERS HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, congenital, megaconial type, 602541

CHM	VISION DISORDERS MENDELIOME	99,2%	98,3%	Choroideremia, 303100
CHMP1A	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	ALS MENDELIOME PARKINSON	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
CHMP4B	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 31, multiple types, 605387
CHN1	VISION DISORDERS MENDELIOME	97,0%	97,0%	Duane retraction syndrome 2, 604356
CHP1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spastic ataxia 9, autosomal recessive, 618438
CHRDL1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Megalocornea 1, X-linked, 309300
CHRM1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CHRM2	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CHRM3	LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Prune belly syndrome, 100100
CHRNA1	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	EPILEPSY MENDELIOME	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, type 4, 610353

CHRNA3	LIVER DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CHRNA4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB1	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	EPILEPSY MENDELIOME	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321
CHRNE	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CHRNG	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS	100,0%	100,0%	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CHST11	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167

CHST14	FETAL AKINESIA (AKI) SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macular corneal dystrophy, 217800
CHST8	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Peeling skin syndrome 3, 616265
CHSY1	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Temptamy preaxial brachydactyly syndrome, 605282
CHUK	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 ?Cocoon syndrome, 613630
CIB1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Epidermodysplasia verruciformis 3, 618267
CIB2	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type II, 614869

FAM92A	MENDELIOME	100,0%	100,0%	?Polydactyly, postaxial, type A9, 618219
CIC	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 45, 617600
CIDEC	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CILK1	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Endocrine-cerebroosteodysplasia, 612651
CISD2	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Wolfram syndrome 2, 604928
CIT	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Filippi syndrome, 272440

CLCC1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 32, 609913
CLCF1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cold-induced sweating syndrome 2, 610313
CLCN1	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive,
CLCN2	MOVEMENT DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN3	INTELLECTUAL DISABILITY MENDELIOME	96,8%	96,7%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Raynaud-Claes syndrome, 300114
CLCN5	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	100,0%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554 Dent disease 1, 300009 Nephrolithiasis, type I, 310468
CLCN6	MENDELIOME	100,0%	100,0%	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173
CLCN7	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLCNKA	MENDELIOME	100,0%	100,0%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090

CLDN1	SKIN DISORDERS LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	HELIX syndrome, 617671
CLDN11	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Leukodystrophy, hypomyelinating, 22, 619328
CLDN14	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 29, 614035
CLDN16	EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypomagnesemia 3, renal, 248250
CLDN19	VISION DISORDERS EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLDN2	MENDELIOME	100,0%	100,0%	?Azoospermia, obstructive, with nephrolithiasis, 301060
CLDN9	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal recessive 116, 619093
CLEC4D	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CLEC7A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Candidiasis, familial, 4, autosomal recessive, 613108
CLIC2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder, X-linked, syndromic 32, 300886

CLIC5	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 103, 616042
CLIP1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
CLMP	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital short bowel syndrome, 615237
CLN3	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	92,7%	92,5%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	71,7%	71,6%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 10, 615803

CLPB	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLPP	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Perrault syndrome 3, 614129
CLPX	MENDELIOME	100,0%	100,0%	?Protoporphyrina, erythropoietic, 2, 618015
CLRN1	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CLRN2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal recessive 117, 619174
CLTC	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 56, 617854
CLTCL1	HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CLUAP1	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CMAS	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CNBP	MENDELIOME	100,0%	100,0%	Myotonic dystrophy 2, 602668

CNGA1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	91,0%	91,0%	Retinitis pigmentosa 49, 613756
CNGA2	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CNGA3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Achromatopsia 2, 216900
CNGB1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 45, 613767
CNGB3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Achromatopsia 3, 262300
CNKS2R2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Houge type, 301008
CNNM2	EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Jalili syndrome, 217080
CNOT1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Vissers-Bodmer syndrome, 619033 Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672

CNP	MENDELIOME	100,0%	100,0%	?Leukodystrophy, hypomyelinating, 20, 619071
CNPY3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 60, 617929
CNTN1	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Myopathy, congenital, Compton-North, 612540
CNTN2	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	FETAL AKINESIA (AKI) NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pitt-Hopkins like syndrome 1, 610042
COA1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COA3	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	85,2%	85,2%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387

COA8	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	93,5%	93,5%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COCH	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 9, 601369 ?Deafness, autosomal recessive 110, 618094
COG1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIg, 611209
COG2	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type III, 613612
COG6	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576

COG7	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIe, 608779
COG8	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 19, 616720
COL14A1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID

COL17A1	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional 4, intermediate, 619787
COL18A1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880
COL1A1	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL1A2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type III, 259420 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210
COL25A1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,9%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Steel syndrome, 615155

	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162 Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloepiphyseal dysplasia, 271700 SMED Strudwick type, 184250 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Kniest dysplasia, 156550 Stickler syndrome, type I, nonsyndromic ocular, 609508 Osteoarthritis with mild chondrodysplasia, 604864 
COL3A1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A1	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COL4A2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Brain small vessel disease 2, 614483
COL4A3	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780

COL4A4	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780
COL4A5	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	100,0%	100,0%	Alport syndrome 1, X-linked, 301050
COL4A6	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, X-linked 6, 300914
COL5A1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Ehlers-Danlos syndrome, classic type, 1, 130000 Fibromuscular dysplasia, multifocal, 619329
COL5A2	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	FETAL AKINESIA (AKI) MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	FETAL AKINESIA (AKI) MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	FETAL AKINESIA (AKI) MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810

COL6A5	NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
COL7A1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, pretibial, 131850 Transient bullous of the newborn, 131705 EBD, Bart type, 132000 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, localisata variant,
COL8A2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969

COLEC10	MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	3MC syndrome 3, 248340
COLEC11	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	3MC syndrome 2, 265050
COLGALT1	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Brain small vessel disease 3, 618360
COLQ	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 5, 603034
COMP	NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
COMT	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
COPA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
COPB1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Baralle-Macken syndrome, 619255
COPB2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 19, primary, autosomal recessive, 617800

COQ2	MOVEMENT DISORDERS EPILEPSY HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	97,2%	97,2%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	HNPD METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 4, 612016

COQ8B	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 9, 615573
COQ9	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	MENDELIOME	100,0%	100,0%	Preeclampsia/eclampsia 5, 614595
CORO1A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 8, 615401
COX10	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	FETAL AKINESIA (AKI) HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 22, 619355

COX20	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COX6A1	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COX6C	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COX7A1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COX7A2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

COX7B	VISION DISORDERS SKIN DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COX7C	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
COX8A	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
CP	MOVEMENT DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPA6	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anterior segment dysgenesis 8, 617319
CPE	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	BDV syndrome, 619326
CPLANE1	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615

CPLX1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 63, 617976
CPN1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Carboxypeptidase N deficiency, 212070
CPOX	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Coproporphyrin, 121300 Harderoporphyrin, 618892
CPS1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Myopia 27, 618827
CPSF3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CPT1A	HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	CPT deficiency, hepatic, type IA, 255120
CPT1C	MENDELIOME	100,0%	100,0%	?Spastic paraparesis 73, autosomal dominant, 616282
CPT2	EPILEPSY HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110

CR2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 7, 614699
CRACR2A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CRADD	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 8, 613835 Retinitis pigmentosa-12, 600105 Pigmented paravenous chorioretinal atrophy, 172870
CRB2	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,1%	96,1%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREB1	MENDELIOME	100,0%	100,0%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XVI, 616229
CREB3L3	MENDELIOME	100,0%	100,0%	Hypertriglyceridemia 2, 619324
CREBBP	DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849

CRELD1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRIPT	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,6%	98,5%	Cold-induced sweating syndrome 1, 272430
CRPPA	FETAL AKINESIA (AKI) METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRTAP	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type VII, 610682
CRTC1	MENDELIOME	100,0%	100,0%	Mucoepidermoid salivary gland carcinoma,
CRX	VISION DISORDERS MENDELIOME	100,0%	100,0%	Leber congenital amaurosis 7, 613829 Cone-rod retinal dystrophy-2, 120970
CRYAA	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 9, multiple types, 604219
CRYAB	VISION DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 10, multiple types, 600881

CRYBA2	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Cataract 42, 115900
CRYBA4	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 23, 610425
CRYBB1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 17, multiple types, 611544
CRYBB2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 3, multiple types, 601547
CRYBB3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 22, 609741
CRYGB	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 2, multiple types, 604307
CRYGD	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 4, multiple types, 115700
CRYGS	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 20, multiple types, 116100
CRYL1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CRYM	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 40, 616357
CSDE1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

CSF1R	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSF2RA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	96,0%	92,4%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830
CSGALNACT1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSNK1D	MENDELIOME	100,0%	100,0%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK1G1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CSNK2A1	INTELLECTUAL DISABILITY MENDELIOME	94,0%	94,0%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Poirier-Bienvenu neurodevelopmental syndrome, 618732

CSPP1	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 21, 615636
CSRP3	HEART MENDELIOME	100,0%	100,0%	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CST3	MENDELIOME	100,0%	100,0%	Cerebral amyloid angiopathy, 105150
CST6	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peeling skin syndrome 4, 607936
CSTB	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	99,4%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 21, 615502

CTDP1	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Cystathioninuria, 219500
CTHRC1	MENDELIOME	100,0%	100,0%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CTNNA1	VISION DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Macular dystrophy, patterned, 2, 608970
CTNNA2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	HEART MENDELIOME	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Exudative vitreoretinopathy 7, 617572 Pilomatrixoma, somatic, 132600 Colorectal cancer, somatic, 114500 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Medulloblastoma, somatic, 155255 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTNNBL1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CTNND1	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Blepharocheilodontic syndrome 2, 617681

CTNND2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CTNS	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTPS1	PRIMARY IMMUNODEFICIENCIES NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	93,0%	93,0%	Immunodeficiency 24, 615897
CTR9	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
CTSA	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galactosialidosis, 256540
CTS B	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CTSC	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CTSD	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 10, 610127

CTSF	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CTSH	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CTSK	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pycnodynostosis, 265800
CTTNBP2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CTU2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Imerslund-Grasbeck syndrome 1, 261100
CUL3	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with or without autism or seizures, 619239 Pseudohypoaldosteronism, type IIE, 614496
CUL4B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Intellectual developmental disorder, X-linked, syndromic, Cabezas type, 300354
CUL7	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	3-M syndrome 1, 273750

CUX1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 67, 618141
CWC27	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCR2	MENDELIOME	100,0%	100,0%	?WHIM syndrome 2, 619407
CXCR4	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYB561	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Orthostatic hypotension 2, 618182
CYB5A	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400

CYBC1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Chronic granulomatous disease 5, autosomal recessive, 618935
CYBRD1	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
CYC1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Thrombocytopenia 4, 612004
CYFIP2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 65, 618008
CYLD	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606 ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
CYP11A1	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110

CYP19A1	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP24A1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypercalcemia, infantile, 1, 143880
CYP26B1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Focal facial dermal dysplasia 4, 614974
CYP27A1	MOVEMENT DISORDERS NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vitamin D-dependent rickets, type I, 264700

CYP2A6	MENDELIOME	100,0%	100,0%	Coumarin resistance, 122700
CYP2B6	MENDELIOME	100,0%	100,0%	Efavirenz, poor metabolism of, 614546
CYP2C19	MENDELIOME	100,0%	100,0%	Proguanil poor metabolizer, 609535 Mephénytoïn poor metabolizer, 609535 Clopidogrel, impaired responsiveness to, 609535 Omeprazole poor metabolizer, 609535
CYP2C8	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
CYP2C9	MENDELIOME	100,0%	100,0%	Warfarin sensitivity, 122700 Tolbutamide poor metabolizer,
CYP2R1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 56, autosomal recessive, 615030
CYP3A4	MENDELIOME	100,0%	99,3%	Vitamin D-dependent rickets, type 3, 619073
CYP4F22	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	MOVEMENT DISORDERS NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812

D2HGDH	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	D-2-hydroxyglutaric aciduria, 600721
DAAM2	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephrotic syndrome, type 24, 619263
DAB1	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 37, 615945
DACT1	MENDELIOME	100,0%	100,0%	?Townes-Brocks syndrome 2, 617466
DAG1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DALRD3	MENDELIOME	100,0%	100,0%	?Developmental and epileptic encephalopathy 86, 618910
DAO	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DARS1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBF4	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

DBH	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBR1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DBT	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Maple syrup urine disease, type II, 248600
DCAF17	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Woodhouse-Sakati syndrome, 241080
DCAF8	NEUROPATHIES MENDELIOME	100,0%	100,0%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Esophageal carcinoma, somatic, 133239 Colorectal cancer, somatic, 114500 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DCDC2	CILIOPATHIES HEARING IMPAIRMENT LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394

DCHS1	HEART INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DCN	VISION DISORDERS MENDELIOME	95,7%	95,7%	Corneal dystrophy, congenital stromal, 610048
DCPS	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Al-Raqad syndrome, 616459
DCT	VISION DISORDERS MENDELIOME	100,0%	100,0%	Oculocutaneous albinism, type VIII, 619165
DCTN1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PARKINSON	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VIIIB, 607641 Perry syndrome, 168605
DCTN2	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DCX	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Subcortical laminar heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DCXR	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DDB1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	White-Kernohan syndrome, 619426

DDB2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	MOVEMENT DISORDERS VISION DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Warburg-Cinotti syndrome, 618175 Spondylometaphyseal dysplasia, short limb-hand type, 271665
DDRGK1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Warsaw breakage syndrome, 613398
DDX23	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

DDX3X	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS	99,2%	97,6%	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX41	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
DDX58	VISION DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Singleton-Merten syndrome 2, 616298
DDX59	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Orofaciodigital syndrome V, 174300
DDX6	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEF6	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 87 and autoimmunity, 619573
DEGS1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 49, 617281

DEPDC5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Epilepsy, familial focal, with variable foci 1, 604364
DES	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGAT1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGAT2	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DGKE	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 7, 615008
DGUOK	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	FETAL AKINESIA (AKI) SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	97,7%	97,7%	Desmosterolosis, 602398

DHCR7	FETAL AKINESIA (AKI) SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	95,2%	95,2%	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DHFR	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Miller syndrome, 263750
DHPS	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	93,2%	93,2%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480

DHTKD1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoacidic and alpha-ketoadipic aciduria, 204750
DHX16	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX37	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 46, XY sex reversal 11, 273250
DHX38	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 84, 618220
DIABLO	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 64, 614152
DIAPH1	HEARING IMPAIRMENT EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	MENDELIOME	100,0%	99,7%	?Premature ovarian failure 2A, 300511
DIAPH3	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Auditory neuropathy, autosomal dominant 1, 609129
DICER1	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295

DIP2B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630
DIS3L2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Perlman syndrome, 267000
DISP1	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DKC1	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Dyskeratosis congenita, X-linked, 305000
DLAT	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Colorectal cancer, somatic, 114500
DLD	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 90, 300850

DLG4	INTELLECTUAL DISABILITY MENDELIOME	98,8%	98,8%	Intellectual developmental disorder, autosomal dominant 62, 618793
DLL1	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLL3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Adams-Oliver syndrome 6, 616589
DLST	MENDELIOME MITOCHONDRIAL DISORDERS HEREDITARY CANCER	100,0%	100,0%	Paragangliomas 7, 618475
DLX3	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	?Orofacial cleft 15, 616788
DLX5	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID

DMAC1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
DMAC2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
DMAC2L	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
DMC1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DMD	HEART INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypophosphatemic rickets, AR, 241520
DMPK	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Myotonic dystrophy 1, 160900
DMRT1	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DMRT2	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DMXL2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113

DNA2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 13, 613193
LRRC6	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 18, 614874
PIH1D3	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spermatogenic failure 18, 617576 ?Ciliary dyskinesia, primary, 37, 617577
DNAH10	NEUROPATHIES MENDELIOME	100,0%	100,0%	Spermatogenic failure 56, 619515

DNAH11	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 39, 618643
DNAH2	MENDELIOME	99,8%	99,3%	Spermatogenic failure 45, 619094
DNAH5	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH8	CILIOPATHIES MENDELIOME	100,0%	100,0%	Spermatogenic failure 46, 619095
DNAH9	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJA3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
DNAJB11	LIVER DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881

DNAJB5	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DNAJB6	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	HEREDITARY BONE MARROW FAILURE SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Bone marrow failure syndrome 3, 617052
DNAJC3	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Leber hereditary optic neuropathy, autosomal recessive, 619382
DNAJC5	EPILEPSY MENDELIOME	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350
DNAJC6	MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528

DNAL1	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 16, 614017
DNAL4	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	?Mirror movements 3, 616059
DNASE1	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DNASE1L3	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Systemic lupus erythematosus 16, 614420
DNASE2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DNHD1	MENDELIOME	100,0%	100,0%	Spermatogenic failure 65, 619712
DNM1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	97,7%	97,4%	Developmental and epileptic encephalopathy 31, 616346
DNM1L	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY NEUROPATHIES HNPD METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	FETAL AKINESIA (AKI) NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368

DNMBP	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 48, 618415
DNMT1	MOVEMENT DISORDERS NEUROPATHIES HNPD METABOLIC DISORDERS MENDELIOME	100,0%	99,7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3A	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Tatton-Brown-Rahman syndrome, 615879 Acute myeloid leukemia, somatic, 601626 Heyn-Sproul-Jackson syndrome, 618724
DNMT3B	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 40, 616433
DOCK3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Adams-Oliver syndrome 2, 614219
DOCK7	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 23, 615859

DOCK8	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Fetal aknesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	SKIN DISORDERS HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type Im, 610768
DONSON	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPCD	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DPF2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 7, 618027
DPH1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901

DPM1	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	99,8%	97,8%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Intellectual developmental disorder, autosomal dominant 33, 616311
DPY19L2	MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spermatogenic failure 9, 613958
DPYD	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dihydropyrimidinuria, 222748

DPYSL5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Ritscher-Schinzel syndrome 4, 619435
DRAM2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 21, 616502
DRC1	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 21, 615294
DRD4	MENDELIOME	100,0%	100,0%	Autonomic nervous system dysfunction,
DRP2	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DSC2	SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis and recurrent skin vesicles, 613102
DSE	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG2	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1BB, 612877 Arrhythmogenic right ventricular dysplasia 10, 610193
DSG3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226

DSG4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis 6, 607903
DSP	SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DSPP	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Dentinogenesis imperfecta, Shields type III, 125500 Dentinogenesis imperfecta, Shields type II, 125490 Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594
DST	SKIN DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	95,6%	95,6%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DSTYK	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNA	HEART MENDELIOME	100,0%	100,0%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 7, 614076
DTYMK	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
DUOX2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid dyshormonogenesis 5, 274900

DUSP6	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Robinow syndrome, autosomal dominant 3, 616894
DYM	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	EPILEPSY NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563
DYNC1I2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091

WDR60	CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYRK1A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 7, 614104
DYRK1B	MENDELIOME	100,0%	100,0%	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768
DZIP1	HEART MENDELIOME	100,0%	100,0%	Spermatogenic failure 47, 619102 ?Mitral valve prolapse 3, 610840
DZIP1L	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polycystic kidney disease 5, 617610

E2F1	MALE INFERTILITY MENDELIOME	100,0%	99,9%	No OMIM Disease ID
EARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	MENDELIOME	90,2%	90,2%	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
ECEL1	FETAL AKINESIA (AKI) SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Arthrogryposis, distal, type 5D, 615065
ECHS1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Urbach-Wiethe disease, 247100
ECSIT	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

EDA	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	100,0%	100,0%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Congenital disorder of glycosylation, type 2V, 619493
EDN1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDN3	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Waardenburg syndrome, type 4B, 613265
EDNRA	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Mandibulofacial dysostosis with alopecia, 616367

EDNRB	SKIN DISORDERS HEARING IMPAIRMENT LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EED	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Cohen-Gibson syndrome, 617561
EEF1A2	EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 33, 616409 Intellectual developmental disorder, autosomal dominant 38, 616393
EEF2	MENDELIOME	100,0%	100,0%	?Spinocerebellar ataxia 26, 609306
EFEMP1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cutis laxa, autosomal recessive, type IB, 614437
EFHC1	EPILEPSY MENDELIOME	98,0%	98,0%	No OMIM Disease ID
EFL1	HEREDITARY BONE MARROW FAILURE SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Shwachman-Diamond syndrome 2, 617941
EFNA4	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

EFNB1	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Craniofrontonasal dysplasia, 304110
EFNB2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
EFTUD2	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Hypomagnesemia 4, renal, 611718
EGFR	MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Non-small cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980
EGLN1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Erythrocytosis, familial, 3, 609820
EGLN2	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
EGR2	FETAL AKINESIA (AKI) NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253
EHD1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
EHHADH	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Fanconi renotubular syndrome 3, 615605

EHMT1	CONGENITAL HEART DISEASE EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,8%	Kleefstra syndrome 1, 610253
EIF2AK1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 Dystonia 33, 619687
EIF2AK3	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Wolcott-Rallison syndrome, 226980
EIF2AK4	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896

EIF2B5	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2S3	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	MEHMO syndrome, 300148
EIF3F	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A3	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF5A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Faundes-Banka syndrome, 619376
ELAC2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 17, 615440
ELANE	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF2	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ELF4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074

ELMO2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vascular malformation, primary intraosseous, 606893
ELMOD3	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 88, 615429 ?Deafness, autosomal dominant 81, 619500
ELN	CONGENITAL HEART DISEASE SKIN DISORDERS HEART MENDELIOME	100,0%	100,0%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	VISION DISORDERS SKIN DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 38, 615957
ELP1	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	Dysautonomia, familial, 223900 Medulloblastoma, 155255
ELP2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 58, 617270
ELP4	MENDELIOME	87,1%	87,1%	?Aniridia 2, 617141

EMC1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EMD	HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bowen-Conradi syndrome, 211180
EMILIN1	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
EML1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Band heterotopia, 600348
EMP2	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 10, 615861
EMX2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Schizencephaly, 269160
EN1	MENDELIOME	100,0%	100,0%	?ENDOVE syndrome, limb-brain type, 619218
ENAM	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 1, 187300

EN03	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease XIII, 612932
ENPP1	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
ENTPD1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	94,3%	90,6%	Adams-Oliver syndrome 4, 615297
EP300	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	MENDELIOME	100,0%	100,0%	Erythrocytosis, familial, 4, 611783
EPB41	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Elliptocytosis-1, 611804
EPB41L1	MENDELIOME	97,8%	97,8%	?Intellectual developmental disorder, autosomal dominant 11, 614257
EPB42	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spherocytosis, type 5, 612690
EPCAM	MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	99,9%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217

EPG5	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Vici syndrome, 242840
EPHA2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 6, multiple types, 116600
EPHA7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
EPHB2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	99,9%	?Bleeding disorder, platelet-type, 22, 618462
EPHB4	MENDELIOME	100,0%	100,0%	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
EPHX1	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
EPHX2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
EPM2A	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
EPRS1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 15, 617951

EPS8	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 102, 615974
EPS8L2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	88,0%	88,0%	Deafness autosomal recessive 106, 617637
EPS8L3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Hypotrichosis 5, 612841
ERAL1	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Perrault syndrome 6, 617565
ERBB2	NEUROPATHIES MENDELIOME	100,0%	100,0%	Gastric cancer, somatic, 613659 Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 ?Visceral neuropathy, familial, 2, autosomal recessive, 619465 Glioblastoma, somatic, 137800
ERBB3	FETAL AKINESIA (AKI) NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Lethal congenital contractual syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERBB4	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756

ERCC3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760 Fanconi anemia, complementation group Q, 615272
ERCC5	FETAL AKINESIA (AKI) SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	FETAL AKINESIA (AKI) SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 ?De Sanctis-Cacchione syndrome, 278800 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946
ERCC6L2	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bone marrow failure syndrome 2, 615715
ERCC8	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400
ERF	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ERGIC1	MENDELIOME PRE CONCEPTION SCREENING	98,4%	98,4%	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100

ERLIN1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 62, 615681
ERLIN2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	MENDELIOME	100,0%	100,0%	?Periventricular nodular heterotopia 6, 615544
ESCO2	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
ESPN	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 Deafness, autosomal recessive 36, 609006 ?Usher syndrome, type 1M, 618632
ESR1	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Breast cancer, somatic, 114480 Estrogen resistance, 615363
ESR2	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY HEREDITARY CANCER	100,0%	100,0%	?Ovarian dysgenesis 8, 618187
ESRP1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 109, 618013
ESRRB	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 35, 608565
ETFA	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glutaric acidemia IIA, 231680

ETFB	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glutaric acidemia IIB, 231680
ETFDH	LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Glutaric acidemia IIC, 231680
ETHE1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Ethylmalonic encephalopathy, 602473
ETV6	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
EVC	CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530

EWSR1	MENDELIOME	100,0%	100,0%	Neuroepithelioma, 612219 Ewing sarcoma, 612219
EXOC2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC6	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
EXOC6B	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepiphyseal dysplasia with joint laxity, type 3, 618395
EXOC7	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOC8	CILIOPATHIES MENDELIOME	100,0%	100,0%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXOSC1	MENDELIOME	100,0%	100,0%	?Pontocerebellar hypoplasia, type 1F, 619304
EXOSC2	VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576

EXOSC8	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
EXT1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	CILIOPATHIES PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425

EYA1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT METABOLIC DISORDERS RENAL DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
EYA4	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 25, 602772
EZH2	HEREDITARY BONE MARROW FAILURE CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Weaver syndrome, 277590
F10	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Factor X deficiency, 227600
F11	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
F13A1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Factor XIII A deficiency, 613225

F13B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Factor XIIIIB deficiency, 613235
F2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia 1 due to thrombin defect, 188050
F2RL3	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
F5	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thrombophilia 2 due to activated protein C resistance, 188055 Factor V deficiency, 227400
F7	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Factor VII deficiency, 227500
F8	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071 Hemophilia A, 306700
F9	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	99,8%	Hemophilia B, 306900 Thrombophilia 8, X-linked, due to factor IX defect, 300807
FA2H	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraparesis 35, autosomal recessive, 612319
FAAH	HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID

FAAP24	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FADD	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAH	HEART LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Tyrosinemia, type I, 276700
FAM111A	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM111B	SKIN DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Joubert syndrome 36, 618763
FAM161A	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 28, 606068
FAM20A	CRANIOFACIAL ANOMALIES SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690

FAM20B	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FAM20C	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Raine syndrome, 259775
FAM50A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261
FAM83G	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FAM83H	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	100,0%	100,0%	Amelogenesis imperfecta, type IIIA, 130900
FAN1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Interstitial nephritis, karyomegalic, 614817
FANCA	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group A, 227650
FANCB	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group B, 300514

FANCC	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	97,3%	97,3%	Fanconi anemia, complementation group C, 227645
FANCD2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	98,8%	98,8%	Fanconi anemia, complementation group D2, 227646
FANCE	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group E, 600901
FANCF	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group F, 603467
FANCG	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group G, 614082
FANCI	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group I, 609053

FANCL	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group L, 614083
FANCM	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY HEREDITARY CANCER	100,0%	100,0%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAR1	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSA	MENDELIOME	100,0%	100,0%	?Rajab interstitial lung disease with brain calcifications 2, 619013
FARSB	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Rajab interstitial lung disease with brain calcifications 1, 613658
FAS	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,

FASLG	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FASTKD2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 44, 618855
FAT1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FAT2	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 45, 617769
FAT4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FBLN5	SKIN DISORDERS NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	91,8%	91,8%	Cutis laxa, autosomal recessive, type IA, 219100 Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 ?Cutis laxa, autosomal dominant 2, 614434
FBN1	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900

FBN2	FETAL AKINESIA (AKI) NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBP1	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	93,7%	93,7%	Fructose-1,6-bisphosphatase deficiency, 229700
FBP2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FBRSL1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,6%	No OMIM Disease ID
FBXL3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO28	MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 100, 619777
FBXO31	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXO32	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FBXO38	NEUROPATHIES MENDELIOME	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IID, 615575

FBXO43	MENDELIOME	100,0%	100,0%	Oocyte maturation defect 12, 619697 Spermatogenic failure 64, 619696
FBXO7	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinson disease 15, autosomal recessive, 260300
FBXW11	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FBXW4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	90,4%	86,2%	No OMIM Disease ID
FBXW7	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
FCGR3A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 20, 615707
FCGR3B	MENDELIOME	99,1%	98,5%	No OMIM Disease ID
FCHO1	PRIMARY IMMUNODEFICIENCIES MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 76, 619164
FCN3	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Squalene synthase deficiency, 618156
FDPS	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Porokeratosis 9, multiple types, 616631

FDX2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Auditory neuropathy and optic atrophy, 617717
FECH	SKIN DISORDERS IRON DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Protoporphryia, erythropoietic, 1, 177000
FERMT1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kindler syndrome, 173650
FERMT3	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypodysfibrinogenemia, congenital, 616004 Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400
FGB	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400

FGD1	CRANIOFACIAL ANOMALIES EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGD4	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 47, 617166
FGF13	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 90, 301058
FGF14	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 27, 609307
FGF16	MENDELIOME	100,0%	100,0%	Metacarpal 4-5 fusion, 309630
FGF17	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Renal hypodysplasia/aplasia 2, 615721

FGF23	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Trichomegaly, 190330
FGF8	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Multiple synostoses syndrome 3, 612961
FGFR1	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Pfeiffer syndrome, 101600 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Hartsfield syndrome, 615465 Trigonocephaly 1, 190440 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

FGFR2	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FGFR3	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 Thanatophoric dysplasia, type II, 187601 Nevus, epidermal, somatic, 162900 CATSHL syndrome, 610474 Thanatophoric dysplasia, type I, 187600 Spermatocytic seminoma, somatic, 273300 Bladder cancer, somatic, 109800 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247
FGG	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400

FH	SKIN DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FHL1	HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myopathy, X-linked, with postural muscle atrophy, 300696 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717
FHL2	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FHOD3	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, familial hypertrophic, 28, 619402
FIBP	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	ALS NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FIGLA	MENDELIOME	100,0%	100,0%	Premature ovarian failure 6, 612310
FIGN	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FITM2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Siddiqi syndrome, 618635

FKBP10	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450
FKBP14	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	FETAL AKINESIA (AKI) HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615
FLAD1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis vulgaris, 146700
FLG2	SKIN DISORDERS MENDELIOME	99,9%	99,9%	Peeling skin syndrome 6, 618084

FLI1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Bleeding disorder, platelet-type, 21, 617443
FLNA	CRANIOFACIAL ANOMALIES EPILEPSY HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FLNB	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FLNC	FETAL AKINESIA (AKI) HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	MENDELIOME	100,0%	100,0%	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	CONGENITAL HEART DISEASE SKIN DISORDERS HEART MENDELIOME	100,0%	100,0%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780

FLVCR1	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	FETAL AKINESIA (AKI) INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FMN2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMO3	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Trimethylaminuria, 602079
FMR1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624 Premature ovarian failure 1, 311360
FN1	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	100,0%	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894
FNIP1	SKIN DISORDERS HEART PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705

FOLR1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXC2	SKIN DISORDERS HEART RENAL DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FOXE1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Bamforth-Lazarus syndrome, 241850
FOXE3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968
FOXF1	CILIOPATHIES MENDELIOME	100,0%	100,0%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXF2	HEARING IMPAIRMENT MENDELIOME	99,0%	97,7%	No OMIM Disease ID
FOXG1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Rett syndrome, congenital variant, 613454

FOXH1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FOXI1	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Enlarged vestibular aqueduct, 600791
FOXJ1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 43, 618699
FOXL1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FOXL2	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Premature ovarian failure 3, 608996
FOXN1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	MENDELIOME	100,0%	100,0%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with language impairment with or without autistic features, 613670
FOXP2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Speech-language disorder-1, 602081
FOXP3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790

FOXRED1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FPR1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FRAS1	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Fraser syndrome 1, 219000
FREM1	VISION DISORDERS SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485
FREM2	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	96,6%	96,6%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	MOVEMENT DISORDERS VISION DISORDERS MENDELIOME	100,0%	99,8%	Nystagmus, infantile periodic alternating, X-linked, 310700 Nystagmus 1, congenital, X-linked, 310700
FRMPD4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	98,3%	98,3%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 37, 616981

FSCN2	MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 30, 607921
FSHB	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300
FSIP2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 34, 618153
FTCD	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glutamate formiminotransferase deficiency, 229100
FTH1	IRON DISORDERS MENDELIOME	100,0%	100,0%	?Hemochromatosis, type 5, 615517
FTL	MOVEMENT DISORDERS VISION DISORDERS IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON	100,0%	100,0%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	94,2%	94,2%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 9, 309549

FUCA1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fucosidosis, 230000
FURIN	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FUS	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FUT6	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FUT8	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FUZ	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
FXN	NEUROPATHIES IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXR1	MENDELIOME	100,0%	100,0%	?Myopathy, congenital, with respiratory insufficiency and bone fractures, 618822 ?Myopathy, congenital proximal, with minicore lesions, 618823
FXYD2	EPILEPSY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypomagnesemia 2, renal, 154020

FYB1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Thrombocytopenia 3, 273900
FYCO1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 18, autosomal recessive, 610019
FZD2	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Omodyplasia 2, 164745
FZD4	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinopathy of prematurity, 133780 Exudative vitreoretinopathy 1, 133780
FZD6	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nail disorder, nonsyndromic congenital, 1, 161050
FZR1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
G6PC	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycogen storage disease Ia, 232200
G6PC3	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Hemolytic anemia, G6PD deficient (favism), 300908

GAA	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease II, 232300
GAB1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 26, 605428
GABBR2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,8%	Developmental and epileptic encephalopathy 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903
GABRA1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 19, 615744
GABRA2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 78, 618557
GABRA3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,8%	No OMIM Disease ID
GABRA5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 43, 617113
GABRD	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

GABRG2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	93,0%	93,0%	Developmental and epileptic encephalopathy 74, 618396 Febrile seizures, familial, 8, 607681 Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 89, 619124
GAL	MENDELIOME	100,0%	100,0%	?Epilepsy, familial temporal lobe, 8, 616461
GALC	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Krabbe disease, 245200
GALE	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galactose epimerase deficiency, 230350
GALK1	VISION DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galactokinase deficiency with cataracts, 230200
GALM	VISION DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Galactosemia IV, 618881
GALNS	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis IVA, 253000
GALNT12	MENDELIOME HEREDITARY CANCER	100,0%	99,8%	No OMIM Disease ID

GALNT2	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type II <sup>t</sup> , 618885
GALNT3	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALNT5	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GALT	VISION DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Galactosemia, 230400
GAMT	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebral creatine deficiency syndrome 2, 612736
GAN	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Giant axonal neuropathy-1, 256850

GANAB	LIVER DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Polycystic kidney disease 3, 600666
GAPVD1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GARS1	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472
GAS2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GAS2L2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 33, 616726
GATA1	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME	100,0%	100,0%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GATA3	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255

GATA4	CONGENITAL HEART DISEASE DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME	100,0%	100,0%	Tetralogy of Fallot, 187500 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430 ?Testicular anomalies with or without congenital heart disease, 615542
GATA5	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Congenital heart defects, multiple types, 5, 617912
GATA6	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrial septal defect 9, 614475 Persistent truncus arteriosus, 217095 Pancreatic agenesis and congenital heart defects, 600001 Atrioventricular septal defect 5, 614474 Tetralogy of Fallot, 187500
GATAD1	HEART MENDELIOME	100,0%	100,0%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	GAND syndrome, 615074
GATB	HEART MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	HEART MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 42, 618839
GATM	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600

GBA	FETAL AKINESIA (AKI) MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Gaucher disease, type II, 230900 Gaucher disease, type IIIC, 231005 Gaucher disease, type III, 231000 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013
GBA2	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	FETAL AKINESIA (AKI) MOVEMENT DISORDERS HEART NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GBF1	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GCDH	MOVEMENT DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glutaricaciduria, type I, 231670
GCGR	MENDELIOME	100,0%	100,0%	Mahvash disease, 619290

GCH1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	EPILEPSY METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	97,0%	93,1%	MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCLM	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GCM2	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343
GCNA	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure, X-linked, 4, 301077
GCNT2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
GCSH	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Glycine encephalopathy, 605899

GDAP1	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, type 4A, 214400
GDAP2	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF11	MENDELIOME	100,0%	99,9%	?Vertebral hypersegmentation and orofacial anomalies, 619122
GDF2	SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia, isolated, with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acromesomelic dysplasia 2A, 200700 Acromesomelic dysplasia 2B, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A2, 112600 ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Brachydactyly, type A1, C, 615072

GDF6	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Leber congenital amaurosis 17, 615360 Multiple synostoses syndrome 4, 617898 Klippel-Feil syndrome 1, autosomal dominant, 118100
GDF9	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	?Premature ovarian failure 14, 618014
GDI1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 41, 300849
GDNF	LIVER DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
GDPD1	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GEMIN4	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GEMIN5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFAP	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Alexander disease, 203450
GFER	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFI1	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107

GFI1B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Bleeding disorder, platelet-type, 17, 187900
GFM1	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFRA1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GGCX	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGPS1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GGT1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Glutathionuria, 231950
GH1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type II, 173100 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type IA, 262400
GHR	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	99,7%	99,7%	Laron dwarfism, 262500 Increased responsiveness to growth hormone, 604271 Growth hormone insensitivity, partial, 604271

GHRHR	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Growth hormone deficiency, isolated partial, 615925
GIGYF1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GIMAP5	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 55, 617827
GINS2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GIPC1	MENDELIOME	100,0%	100,0%	Oculopharyngodistal myopathy 2, 618940
GIPC3	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 15, 601869
GJA1	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Erythrokeratoderma variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJA3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 14, multiple types, 601885

GJA5	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 1, multiple types, 116200
GJB1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Keratoderma, palmoplantar, with deafness, 148350 Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Hystrix-like ichthyosis with deafness, 602540 Bart-Pumphrey syndrome, 149200 Keratitis-ichthyosis-deafness syndrome, 148210 Vohwinkel syndrome, 124500
GJB3	SKIN DISORDERS HEARING IMPAIRMENT NEUROPATHIES MENDELIOME	100,0%	100,0%	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,
GJB4	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290
GJC2	MOVEMENT DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,5%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804

GK	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Glycerol kinase deficiency, 307030
GLA	SKIN DISORDERS HEARING IMPAIRMENT HEART NEUROPATHIES HNPD METABOLIC DISORDERS RENAL DISORDERS MENDELIOME	91,3%	91,3%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	MOVEMENT DISORDERS SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycine encephalopathy, 605899
GLDN	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lethal congenital contracture syndrome 11, 617194
GLE1	FETAL AKINESIA (AKI) NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GLI1	MENDELIOME	100,0%	100,0%	Polydactyly, preaxial I, 174400 Polydactyly, postaxial, type A8, 618123

GLI2	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Greig cephalopolysyndactyl syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GLIS2	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 7, 611498
GLIS3	LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	Skin Disorders MENDELIOME	100,0%	100,0%	Glomuvenous malformations, 138000
GLRA1	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperekplexia 1, 149400
GLRB	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperekplexia 2, 614619

GLRX5	IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Developmental and epileptic encephalopathy 71, 618328
GLUD1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glutamine deficiency, congenital, 610015
GLYCTK	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	D-glyceric aciduria, 220120
GLYR1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GM2A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	GM2-gangliosidosis, AB variant, 272750
GMNN	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Meier-Gorlin syndrome 6, 616835

GMPPA	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	FETAL AKINESIA (AKI) HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GMPR	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
GMPS	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GNA11	SKIN DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNA14	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GNA1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GNA2	MENDELIOME	100,0%	100,0%	Ventricular tachycardia, idiopathic, 192605 Pituitary adenoma, ACTH-secreting, somatic,
GNA3	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Auriculocondylar syndrome 1, 602483
GNAL	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Dystonia 25, 615073

GNAO1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	83,9%	82,0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ia, 103580 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 McCune-Albright syndrome, somatic, mosaic, 174800 Pseudopseudohypoparathyroidism, 612463
GNAS-AS1	MENDELIOME	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Achromatopsia 4, 613856
GNB1	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286 Leukemia, acute lymphoblastic, somatic, 613065 Intellectual developmental disorder, autosomal dominant 42, 616973
GNB2	HEART MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 ?Sick sinus syndrome 4, 619464
GNB3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary, type 1H, 617024
GNB4	NEUROPATHIES MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 Intellectual developmental disorder with cardiac arrhythmia, 617173

GNE	HEMOSTATIC/THROMBOTIC DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycine N-methyltransferase deficiency, 606664
GNPAT	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	?Rhizomelic dysplasia, Ain-Naz type, 616510
GNPTAB	CRANIOFACIAL ANOMALIES HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucolipidosis III gamma, 252605

GNRH1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GON7	MENDELIOME	100,0%	100,0%	Galloway-Mowat syndrome 9, 619603
GORAB	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Geroderma osteodysplasticum, 231070
GOSR2	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 6, 614018
GOT1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Developmental and epileptic encephalopathy 82, 618721

GP1BA	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,0%	96,3%	Bleeding disorder, platelet-type, 11, 614201
GP9	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bernard-Soulier syndrome, type C, 231200
GPAA1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	99,9%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Keipert syndrome, 301026
GPC6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Omodysplasia 1, 258315

GPD1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	HEART METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Brugada syndrome 2, 611777
GPHN	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Molybdenum cofactor deficiency C, 615501
GPI	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	95,5%	95,5%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	MENDELIOME	100,0%	100,0%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS MENDELIOME	100,0%	100,0%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR161	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
GPR179	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565

GPR68	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, X-linked 7, 301018
GPSM2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chudley-McCullough syndrome, 604213
GPT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GPX4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 114, 618456
GREB1L	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 80, 619274 Renal hypodysplasia/aplasia 3, 617805
GREM1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
GREM2	MENDELIOME	100,0%	100,0%	Tooth agenesis, selective, 9, 617275

GRHL2	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GRHL3	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	van der Woude syndrome 2, 606713
GRHPR	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Hyperoxaluria, primary, type II, 260000
GRIA2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,7%	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIA4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	96,3%	96,3%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 Intellectual developmental disorder, autosomal recessive 6, 611092

GRIN1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Developmental and epileptic encephalopathy 101, 619814 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 27, 616139 Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,3%	Developmental and epileptic encephalopathy 46, 617162
GRIP1	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fraser syndrome 3, 617667
GRK1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Oguchi disease-2, 613411
GRM1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 13, 614831 Spinocerebellar ataxia 44, 617691
GRM6	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRM7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922

GRN	ALS MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 25, 613285
GRXCR2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 101, 615837
GSC	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSDME	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 5, 600994
GSE1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
GSN	VISION DISORDERS SKIN DISORDERS NEUROPATHIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	Amyloidosis, Finnish type, 105120
GSR	MENDELIOME	100,0%	100,0%	Hemolytic anemia due to glutathione reductase deficiency, 618660
GSS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GSX2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646

GTF2E2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	72,5%	72,5%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Jaber-Elahi syndrome, 617988
GTPBP3	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 48, 613827
GUCY1A1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Moyamoya 6 with achalasia, 615750
GUCY2C	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 6, 601777 ?Choroidal dystrophy, central areolar 1, 215500 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1l, 618555
GUF1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Developmental and epileptic encephalopathy 40, 617065
GULOP	MENDELIOME	NC	NC	Scurvy,

GUSB	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis VII, 253220
GYG1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease 0, muscle, 611556
GYS2	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycogen storage disease 0, liver, 240600
GZF1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joint laxity, short stature, and myopia, 617662
H1-4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Rahman syndrome, 617537
H19	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	NC	NC	No OMIM Disease ID
H3-3A	MENDELIOME	100,0%	100,0%	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720
H3-3B	MENDELIOME	100,0%	100,0%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721
H4C11	MENDELIOME	100,0%	100,0%	?Tessadori-van Haaften neurodevelopmental syndrome 2, 619759
H4C3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Tessadori-van Haaften neurodevelopmental syndrome 1, 619758

H6PD	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cortisone reductase deficiency 1, 604931
HAAO	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HABP2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HACE1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	HEART NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	HEART NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Trifunctional protein deficiency, 609015

HAGH	METABOLIC DISORDERS MENDELIOME	100,0%	99,7%	No OMIM Disease ID
HAMP	IRON DISORDERS LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemochromatosis, type 2B, 613313
HAND1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HAND2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HARS1	VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Perrault syndrome 2, 614926
HAVCR2	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Neutropenia, severe congenital 3, autosomal recessive, 610738

HBA1	MENDELIOME	100,0%	100,0%	Hemoglobin H disease, nondeletional, 613978 Thalassemias, alpha-, 604131 Heinz body anemias, alpha-, 140700 Methemoglobinemia, alpha type, 617973 Erythrocytosis 7, 617981
HBA2	MENDELIOME	100,0%	100,0%	Heinz body anemia, 140700 Erythrocytosis 7, 617981 Thalassemia, alpha-, 604131 Hemoglobin H disease, deletional and nondeletional, 613978
HBB	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methemoglobinemia, beta type, 617971 Thalassemia-beta, dominant inclusion-body, 603902 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Delta-beta thalassemia, 141749 Hereditary persistence of fetal hemoglobin, 141749 Heinz body anemia, 140700 Erythrocytosis 6, 617980
HBD	MENDELIOME	100,0%	100,0%	Thalassemia due to Hb Lepore, Thalassemia, delta-,
HBG1	MENDELIOME	98,5%	97,9%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	MENDELIOME	100,0%	100,0%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HCN1	EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME	98,5%	98,5%	Developmental and epileptic encephalopathy 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482

HCN2	HEART HNPD MENDELIOME	94,1%	91,3%	Febrile seizures, familial, 2, 602477 Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	HEART HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HCN4	HEART MENDELIOME	100,0%	100,0%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HCRT	MENDELIOME	100,0%	100,0%	?Narcolepsy 1, 161400
HDAC4	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC6	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	96,6%	96,0%	Cornelia de Lange syndrome 5, 300882
HEATR5B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HECW2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911

HEPACAM	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPH	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HEPHL1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 38, 615516
HES7	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sandhoff disease, infantile, juvenile, and adult forms, 268800

HEY2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HFE	HEART IRON DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Hemochromatosis, 235200
HFM1	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Premature ovarian failure 9, 615724
HGD	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alkaptonuria, 203500
HGF	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 39, 608265
HGSNAT	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	92,1%	92,1%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HHAT	MENDELIOME	100,0%	100,0%	Nivelon-Nivelon-Mabille syndrome, 600092
HIBADH	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HIBCH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620

HID1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HIKESHI	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 43, 616977
HJV	HEART IRON DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemochromatosis, type 2A, 602390
HK1	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HKDC1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 92, 619614
HLCS	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Holocarboxylase synthetase deficiency, 253270

HMBS	SKIN DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HMGA2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	90,0%	81,8%	Silver-Russell syndrome 5, 618908
HMGB3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Microphthalmia, syndromic 13, 300915
HMGCL	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	HMG-CoA lyase deficiency, 246450
HMGCS2	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	IRON DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Heme oxygenase-1 deficiency, 614034
HMX1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Oculoauricular syndrome, 612109
HNF1A	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700
HNF1B	LIVER DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920

HNF4A	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850
HNMT	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 51, 616739
HNRNPA1	MENDELIOME	100,0%	100,0%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426
HNRNPA2B1	MENDELIOME	100,0%	100,0%	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPD	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HNRNPDL	MENDELIOME	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HNRNPH2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Bain type, 300986
HNRNPK	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Au-Kline syndrome, 616580
HNRNPU	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 54, 617391
HOGA1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperoxaluria, primary, type III, 613616
HOMER2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 68, 616707
HOXA1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536

HOXA11	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	99,7%	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305
HOXA2	CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Facial paresis, hereditary congenital, 3, 614744
HOXB13	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
HOXC13	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	NEUROPATHIES MENDELIOME	100,0%	100,0%	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950
HOXD13	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Syndactyly, type V, 186300 Synpolydactyly 1, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 ?Brachydactyly-syndactyly syndrome, 610713
HPCA	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710

HPDL	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HPGD	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranoosteopathia, 259100
HPRT1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HPS1	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 1, 203300
HPS3	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 3, 614072
HPS4	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 4, 614073

HP55	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 5, 614074
HPS6	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Urofacial syndrome 1, 236730
HR	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Atrichia with papular lesions, 209500 Alopecia universalis, 203655
HRAS	VISION DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Congenital myopathy with excess of muscle spindles, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HRG	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Thrombophilia 11 due to HRG deficiency, 613116
HS2ST1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HS3ST6	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	99,6%	?Angioedema, hereditary, 8, 619367

HS6ST1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HS6ST2	MENDELIOME	100,0%	100,0%	?Paganini-Miozzo syndrome, 301025
HSCB	IRON DISORDERS MENDELIOME	100,0%	100,0%	?Anemia, sideroblastic, 5, 619523
HSD11B1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Cortisone reductase deficiency 2, 614662
HSD11B2	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Apparent mineralocorticoid excess, 218030
HSD17B10	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	HSD10 mitochondrial disease, 300438
HSD17B3	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,0%	99,0%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	MOVEMENT DISORDERS HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400

HSD3B2	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	LIVER DISORDERS METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bile acid synthesis defect, congenital, 1, 607765
HSF2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HSF2BP	MENDELIOME	100,0%	100,0%	Premature ovarian failure 19, 619245
HSF4	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 5, multiple types, 116800
HSPA9	IRON DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPB1	NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	NEUROPATHIES MENDELIOME	100,0%	100,0%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB6	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HSPB8	NEUROPATHIES MENDELIOME	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673

HSPD1	MOVEMENT DISORDERS HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	MENDELIOME	100,0%	100,0%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	90,4%	90,3%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779
HTRA2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	3-methylglutaconic aciduria, type VIII, 617248
HTT	MENDELIOME	100,0%	100,0%	Lopes-Macié-Rodan syndrome, 617435 Huntington disease, 143100
HUWE1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, Turner type, 309590
HYAL1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis type IX, 601492
HYAL2	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
HYDIN	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 5, 608647

HYLS1	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Hydrocephalus syndrome, 236680
HYOU1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency 59 and hypoglycemia, 233600
IARS1	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraparesis 74, autosomal recessive, 616451
ICOS	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 1, 607594
ICOSLG	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ID4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IDH1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID

IDH2	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3A	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 90, 619007
IDH3B	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 46, 612572
IDI1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IDS	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Mucopolysaccharidosis II, 309900
IDUA	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis IIs, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IER3IP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, epilepsy, and diabetes syndrome, 614231

IFIH1	EPILEPSY PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Osteogenesis imperfecta, type V, 610967
IFNAR1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	97,8%	97,8%	No OMIM Disease ID
IFNAR2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Immunodeficiency 45, 616669
IFNG	PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	?Immunodeficiency 69, mycobacteriosis, 618963
IFngr1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFngr2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 28, mycobacteriosis, 614889
IFNLR1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IFRD1	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

IFT122	CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cranoectodermal dysplasia 1, 218330
IFT140	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 19, 615996

IFT43	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	?Orofaciodigital syndrome XVIII, 617927
IFT74	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582 ?Bardet-Biedl syndrome 22, 617119
IFT80	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	95,0%	95,0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895

IFT88	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IGBP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Insulin-like growth factor I, resistance to, 270450
IGF2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Silver-Russell syndrome 3, 616489
IGF2R	MENDELIOME	100,0%	100,0%	Hepatocellular carcinoma, somatic, 114550
IGFALS	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acid-labile subunit, deficiency of, 615961
IGFBP7	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	MENDELIOME	100,0%	100,0%	IgG2 deficiency, selective,
IGHM	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Agammaglobulinemia 1, 601495
IGHMBP2	FETAL AKINESIA (AKI) NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155

IGKC	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kappa light chain deficiency, 614102
IGLL1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Agammaglobulinemia 2, 613500
IGSF1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF10	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IGSF3	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Lacrimal duct defect, 149700
IHH	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636

IKZF1	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Immunodeficiency, common variable, 13, 616873
IKZF3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency 84, 619437
IKZF5	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Thrombocytopenia, autosomal dominant, 7, 619130
IL10	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IL10RA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Craniosynostosis and dental anomalies, 614188
IL12B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	94,1%	94,1%	Immunodeficiency 30, 614891
IL17F	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 51, 613953

IL17RC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Candidiasis, familial, 9, 616445
IL17RD	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL18BP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IL1RAPL1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 21, 300143
IL1RN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Interleukin 1 receptor antagonist deficiency, 612852
IL2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IL21	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency, common variable, 11, 615767
IL21R	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 56, 615207
IL2RA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495

IL2RG	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Psoriasis 14, pustular, 614204
IL37	MENDELIOME	100,0%	100,0%	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398
IL6R	PRIMARY IMMUNODEFICIENCIES MENDELIOME	92,7%	92,7%	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Stuve-Wiedemann syndrome 2, 619751 Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752 ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523
IL7R	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 42, 609646
ILK	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IMPA1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 59, 617323

IMPDH1	VISION DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Macular dystrophy, vitelliform, 4, 616151 Retinitis pigmentosa 91, 153870
IMPG2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152
INF2	NEUROPATHIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	MENDELIOME	100,0%	100,0%	Squamous cell carcinoma, head and neck, somatic, 275355
INO80	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
INPP5E	VISION DISORDERS CILIOPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Opsismodysplasia, 258480
INS	MENDELIOME	100,0%	100,0%	Diabetes mellitus, insulin-dependent, 2, 125852 Maturity-onset diabetes of the young, type 10, 613370 Hyperproinsulinemia, 616214 Diabetes mellitus, permanent neonatal 4, 618858

INSL3	MENDELIOME	80,7%	80,7%	Cryptorchidism, 219050
INSR	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INTS1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS8	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	CRANIOFACIAL ANOMALIES CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	VISION DISORDERS CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 2, infantile, 602088
IPMK	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
IPO8	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	VISS syndrome, 619472
IQCB1	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Senior-Loken syndrome 5, 609254

IQCE	MENDELIOME	100,0%	100,0%	Polydactyly, postaxial, type A7, 617642
IQSEC1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,6%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99,8%	99,1%	Intellectual developmental disorder, X-linked 1, 309530
IRAK1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IRAK4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 67, 607676
IРЕB2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF1	MENDELIOME	100,0%	100,0%	Nonsmall cell lung cancer, somatic, 211980 Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic, Myelogenous leukemia, acute,
IRF2BP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IRF4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

IRF6	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MENDELIOME SCHISIS	100,0%	100,0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome 1, 119300
IRF7	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Immunodeficiency 39, 616345
IRF8	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IRS4	MENDELIOME	100,0%	100,0%	Hypothyroidism, congenital, nongoitrous, 9, 301035
IRX1	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IRX5	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hamamy syndrome, 611174
ISCA1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	95,1%	95,1%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myopathy with lactic acidosis, hereditary, 255125

ISG15	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 38, 616126
ITCH	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	95,6%	93,9%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ITGA2B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glanzmann thrombasthenia 1, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related,
ITGA3	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA6	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817
ITGA7	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	97,2%	97,2%	Leukocyte adhesion deficiency, 116920
ITGB3	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 Glanzmann thrombasthenia 2, 619267 Thrombocytopenia, neonatal alloimmune, Purpura, posttransfusion,

ITGB4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 Epidermolysis bullosa, junctional 5A, intermediate, 619816
ITGB6	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IH, 616221
ITK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Lymphoproliferative syndrome 1, 613011
ITM2B	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPA	EPILEPSY HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 35, 616647
ITPKB	PRIMARY IMMUNODEFICIENCIES MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	No OMIM Disease ID
ITPR1	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
ITPR2	MENDELIOME	100,0%	100,0%	?Anhidrosis, isolated, with normal sweat glands, 106190
ITPR3	NEUROPATHIES PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ITSN1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID

ITSN2	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
IVD	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Isovaleric acidemia, 243500
IVNS1ABP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 70, 618969
IYD	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid dyshormonogenesis 4, 274800
JAG1	VISION DISORDERS CONGENITAL HEART DISEASE HEART NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAG2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,6%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAGN1	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Autoinflammation, immune dysregulation, and eosinophilia, 618999

JAK2	HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
JAK3	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM2	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	92,3%	92,3%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JARID2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
JMJD1C	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
JPH1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 2E, 619492 Cardiomyopathy, hypertrophic, 17, 613873
JPH3	MENDELIOME	100,0%	100,0%	Huntington disease-like 2, 606438
JUP	SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528

KALRN	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
KANK1	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Koolen-De Vries syndrome, 610443
KARS1	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 89, 613916 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
CCDC155	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KAT5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Arboleda-Tham syndrome, 616268
KAT6B	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KAT8	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974

KATNB1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lissencephaly 6, with microcephaly, 616212
KIAA0556	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 26, 616784
KBTBD13	NEUROPATHIES MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	MOVEMENT DISORDERS EPILEPSY MENDELIOME	100,0%	100,0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 32, 616366
KCNA4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNAS5	HEART MENDELIOME	100,0%	100,0%	Atrial fibrillation, familial, 7, 612240
KCNB1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Epilepsy, progressive myoclonic 7, 616187

KCNC3	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	99,8%	98,8%	Spinocerebellar ataxia 13, 605259
KCNND2	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KCNND3	MOVEMENT DISORDERS HEART MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399
KCNE1	HEARING IMPAIRMENT HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	HEART MENDELIOME	100,0%	100,0%	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493
KCNE3	HEART MENDELIOME	100,0%	100,0%	?Brugada syndrome 6, 613119
KCNE4	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KCNE5	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KCNH1	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	98,7%	98,7%	Zimmermann-Laband syndrome 1, 135500 Temple-Baraitser syndrome, 611816
KCNH2	HEART MENDELIOME	100,0%	100,0%	Short QT syndrome 1, 609620 Long QT syndrome 2, 613688
KCNJ1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bartter syndrome, type 2, 241200

KCNJ10	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	EPILEPSY HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186
KCNJ16	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Hypokalemic tubulopathy and deafness, 619406
KCNJ2	HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME  SCHISIS MUSCLE DISORDERS	100,0%	100,0%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KCNJ5	HEART RENAL DISORDERS MENDELIOME	100,0%	100,0%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ6	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Keppen-Lubinsky syndrome, 614098
KCNJ8	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID

KCNK3	HEART MENDELIOME	100,0%	100,0%	Pulmonary hypertension, primary, 4, 615344
KCNK4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 6:
KCNK9	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	97,3%	97,3%	Birk-Barel syndrome, 612292
KCNMA1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729
KCNN2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Dystonia 34, myoclonic, 619724 Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725
KCNN3	HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Zimmermann-Laband syndrome 3, 618658
KCNN4	MENDELIOME	100,0%	100,0%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	HEARING IMPAIRMENT HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
KCNQ1OT1	MENDELIOME	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200

KCNQ3	EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Seizures, benign neonatal, 2, 121201
KCNQ4	HEARING IMPAIRMENT MENDELIOME	99,9%	99,3%	Deafness, autosomal dominant 2A, 600101
KCNQ5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 46, 617601
KCNT1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,6%	Developmental and epileptic encephalopathy 14, 614959 Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 57, 617771
KCNV2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinal cone dystrophy 3B, 610356
KCTD1	MENDELIOME	100,0%	100,0%	Scalp-ear-nipple syndrome, 181270
KCTD17	MENDELIOME	100,0%	100,0%	Dystonia 26, myoclonic, 616398
KCTD3	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KCTD7	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDELR2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Osteogenesis imperfecta, type XXI, 619131

KDF1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Diets-Jongmans syndrome, 618846
KDM4B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 65, 619320
KDM5B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	95,6%	94,1%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDM5C	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6A	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Kabuki syndrome 2, 300867
KDM6B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KDR	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Hemangioma, capillary infantile, somatic, 602089
KDSR	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Erythrokeratoderma variabilis et progressiva 4, 617526

KERA	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hydatidiform mole, recurrent, 2, 614293
KHK	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KIAA0586	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	95,8%	95,8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	MENDELIOME	100,0%	100,0%	Polydactyly, postaxial, type A10, 618498
KIAA1109	FETAL AKINESIA (AKI) INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,7%	Retinitis pigmentosa 86, 618613
KIDINS220	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 Ventriculomegaly and arthrogryposis, 619501
KIF11	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950

KIF12	MENDELIOME	100,0%	100,0%	Cholestasis, progressive familial intrahepatic, 8, 619662
KIF14	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	MOVEMENT DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	98,0%	98,0%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	NEUROPATHIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Pheochromocytoma, 171300 Charcot-Marie-Tooth disease, type 2A1, 118210
KIF1C	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic ataxia 2, autosomal recessive, 611302
KIF20A	MENDELIOME	100,0%	100,0%	?Cardiomyopathy, familial restrictive, 6, 619433
KIF21A	VISION DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 3B, 135700 Fibrosis of extraocular muscles, congenital, 1, 135700
KIF21B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KIF22	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Spondyloepiphyseal dysplasia with joint laxity, type 2, 603546
KIF23	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME	100,0%	100,0%	Anemia, congenital dyserythropoietic, type IIIA, 105600

KIF2A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 89, 618955
KIF4A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder, X-linked 100, 300923
KIF5A	ALS MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	FETAL AKINESIA (AKI) INTELLECTUAL DISABILITY MENDELIOME	99,8%	99,8%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrocephalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	96,1%	96,1%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephrotic syndrome, type 23, 619201

KIRREL3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KISS1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIT	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Piebaldism, 172800 Germ cell tumors, somatic, 273300 Mastocytosis, systemic, somatic, 154800 Leukemia, acute myeloid, somatic, 601626
KITLG	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
KIZ	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 69, 615780
KL	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,8%	99,4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLB	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KLC2	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraparesis, optic atrophy, and neuropathy, 609541

KLF1	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME	100,0%	100,0%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
KLF11	MENDELIOME	100,0%	100,0%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	MENDELIOME	100,0%	100,0%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLF7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KLHL10	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 11, 615081
KLHL15	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 103, 300982
KLHL24	SKIN DISORDERS HEART MENDELIOME	100,0%	100,0%	Epidermolysis bullosa simplex 6, generalized, with scarring and hair loss, 617294
KLHL3	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Nemaline myopathy 9, 615731
KLHL7	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KLHL9	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	No OMIM Disease ID

KLK4	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Cowden syndrome 4, 615107
KMT2A	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Wiedemann-Steiner syndrome, 605130
KMT2B	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,7%	99,3%	Dystonia 28, childhood-onset, 617284
KMT2C	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Kleefstra syndrome 2, 617768
KMT2D	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE SKIN DISORDERS HEART PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Kabuki syndrome 1, 147920
KMT2E	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	O'Donnell-Luria-Rodan syndrome, 618512

KMT5B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 51, 617788
KNG1	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Angioedema, hereditary, 6, 619363
KNL1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	98,9%	98,9%	Microcephaly 4, primary, autosomal recessive, 604321
KNSTRN	MENDELIOME	100,0%	100,0%	?Roifman-Chitayat syndrome, digenic, 613328
KPTN	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 41, 615637
KRAS	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
KREMEN1	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	MENDELIOME	100,0%	100,0%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 Cerebral cavernous malformations-1, 116860 Cavernous malformations of CNS and retina, 116860

KRT1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Epidermolytic hyperkeratosis, 113800 Palmoplantar keratoderma, nonepidermolytic, 600962 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Ichthyosis histrix, Curth-Macklin type, 146590
KRT10	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	VISION DISORDERS MENDELIOME	100,0%	100,0%	Meesmann corneal dystrophy 1, 122100
KRT13	SKIN DISORDERS MENDELIOME	100,0%	100,0%	White sponge nevus 2, 615785
KRT14	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 Epidermolysis bullosa simplex 1C, localized, 131800 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex 1A, generalized severe, 131760 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex 1B, generalized intermediate, 131900
KRT16	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Steatocystoma multiplex, 184500 Pachyonychia congenita 2, 167210
KRT18	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cirrhosis, cryptogenic, 215600
KRT2	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Ichthyosis bullosa of Siemens, 146800
KRT25	MENDELIOME	100,0%	100,0%	Woolly hair, autosomal recessive 3, 616760
KRT3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Meesmann corneal dystrophy 2, 618767

KRT4	SKIN DISORDERS MENDELIOME	100,0%	100,0%	White sponge nevus 1, 193900
KRT5	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa simplex 2A, generalized severe, 619555 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 Epidermolysis bullosa simplex 2C, localized, 619594 Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352
KRT6A	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Pachyonychia congenita 3, 615726
KRT6B	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Pachyonychia congenita 4, 615728
KRT6C	SKIN DISORDERS MENDELIOME	100,0%	99,9%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Hypotrichosis 13, 615896
KRT74	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981 ?Ectodermal dysplasia 7, hair/nail type, 614929
KRT75	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
KRT8	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cirrhosis, cryptogenic, 215600
KRT81	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Monilethrix, 158000
KRT83	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Monilethrix, 158000 Erythrokeratoderma variabilis et progressiva 5, 617756
KRT85	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia 4, hair/nail type, 602032

KRT86	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Monilethrix, 158000
KRT9	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Palmoplantar keratoderma, epidermolytic, 144200
KY	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, myofibrillar, 7, 617114
KYNU	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L1CAM	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	MASA syndrome, 303350 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus due to aqueductal stenosis, 307000
L2HGDH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	L-2-hydroxyglutaric aciduria, 236792
LACC1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Juvenile arthritis, 618795
LACTB	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
LAGE3	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Poretti-Boltshauser syndrome, 615960

LAMA2	HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, junctional 2A, intermediate, 619783 Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 Epidermolysis bullosa, junctional 2B, severe, 619784
LAMA4	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1JJ, 615235
LAMA5	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LAMB1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lissencephaly 5, 615191
LAMB2	VISION DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, junctional 1B, severe, 226700 Epidermolysis bullosa, junctional 1A, intermediate, 226650 Amelogenesis imperfecta, type IA, 104530
LAMC2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermolysis bullosa, junctional 3B, severe, 619786 Epidermolysis bullosa, junctional 3A, intermediate, 619785
LAMC3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cortical malformations, occipital, 614115

LAMP2	VISION DISORDERS HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Danon disease, 300257
LAMTOR2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAPTM5	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LARGE1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alazami syndrome, 615071
LARS1	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Infantile liver failure syndrome 1, 615438
LARS2	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Wilson-Turner syndrome, 309585

LAT	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 52, 617514
LBR	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471 Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 Greenberg skeletal dysplasia, 215140
LBX1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	?Central hypoventilation syndrome, congenital, 3, 619483
LCA5	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 5, 604537
LCAT	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fish-eye disease, 136120 Norum disease, 245900
LCK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	?Immunodeficiency 22, 615758
LCP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	?Immunodeficiency 81, 619374
LCT	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lactase deficiency, congenital, 223000
LDB3	HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493

LDHA	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease XI, 612933
LDHB	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LDHD	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	D-lactic aciduria with susceptibility to gout, 245450
LDLR	MENDELIOME	100,0%	100,0%	LDL cholesterol level QTL2, 143890 Hypercholesterolemia, familial, 1, 143890
LDLRAP1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypercholesterolemia, familial, 4, 603813
LEF1	MENDELIOME	100,0%	100,0%	Sebaceous tumors, somatic,
LEFTY2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LEMD2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Marbach-Rustad progeroid syndrome, 619322 Cataract 46, juvenile-onset, 212500
LEMD3	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LEP	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Obesity, morbid, due to leptin deficiency, 614962

LEPR	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	94,6%	94,6%	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,3%	96,6%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	EPILEPSY MENDELIOME	100,0%	100,0%	Epilepsy, familial temporal lobe, 1, 600512
LGI4	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LGR4	MENDELIOME	100,0%	100,0%	Delayed puberty, self-limited, 619613
LHB	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 67, 610265
LHX1	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID

LHX3	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 96, 619774
LIG3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIG4	HEREDITARY BONE MARROW FAILURE DKC PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID) HEREDITARY CANCER	100,0%	100,0%	LIG4 syndrome, 606593

LIM2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 19, multiple types, 615277
LIMS2	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 64, 618103
LINS1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 27, 614340
LIPA	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	95,2%	95,2%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Hepatic lipase deficiency, 614025
LIPE	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipodystrophy, familial partial, type 6, 615980
LIPH	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Lipoyltransferase 1 deficiency, 616299
LIPT2	EPILEPSY METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668

LITAF	NEUROPATHIES MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Combined factor V and VIII deficiency, 227300
LMAN2L	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Intellectual developmental disorder, autosomal recessive 52, 616887
LMBR1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	98,7%	98,7%	Triphalangeal thumb, type I, 174500 Syndactyly, type IV, 186200 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500 Acheiropody, 200500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,1%	96,1%	Methylmalonic aciduria and homocystinuria, cbIF type, 277380
LMBRD2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental delay with variable neurologic and brain abnormalities, 619694
LMCD1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LMF1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipase deficiency, combined, 246650

LMNA	FETAL AKINESIA (AKI) SKIN DISORDERS HEART NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMNB1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Leukodystrophy, adult-onset, autosomal dominant, 169500 Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LMOD1	MENDELIOME	100,0%	100,0%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LMOD3	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Nemaline myopathy 10, 616165
LMX1A	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 7, 601412
LMX1B	VISION DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	100,0%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LNPK	MENDELIOME PRE CONCEPTION SCREENING	93,3%	93,3%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090

LONP1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	CODAS syndrome, 600373
LORICRIN	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Vohwinkel syndrome with ichthyosis, 604117
LOX	MENDELIOME	100,0%	100,0%	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 77, 613079
LOXL3	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Myopia 28, autosomal recessive, 619781
LPAR6	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Majeed syndrome, 609628

LPL	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LPP	MENDELIOME	100,0%	100,0%	Leukemia, acute myeloid, 601626 Lipoma,
LRAT	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIF1	MENDELIOME	100,0%	100,0%	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477
LRIG2	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Urofacial syndrome 2, 615112
LRIG3	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LRIT3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,6%	99,6%	Albinism, oculocutaneous, type VII, 615179
LRP1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Keratosis pilaris atrophicans, 604093
LRP12	MENDELIOME	100,0%	100,0%	Oculopharyngodistal myopathy 1, 164310

LRP2	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Donnai-Barrow syndrome, 222448
LRP4	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	VISION DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
LRP6	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	Tooth agenesis, selective, 7, 616724
LRPAP1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopia 23, autosomal recessive, 615431
LRPPRC	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC10	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
LRRC32	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC56	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 39, 618254

LRRC8A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Agammaglobulinemia 5, 613506
LRRK1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Osteosclerotic metaphyseal dysplasia, 615198
LRRK2	MENDELIOME PARKINSON	100,0%	100,0%	No OMIM Disease ID
LRSAM1	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 63, 611451
LSM11	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Aicardi-Goutieres syndrome 8, 619486
LSS	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-intellectual disability syndrome 4, 618840
LTBP1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 ?Weill-Marchesani syndrome 3, recessive, 614819

LTPB3	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTPB4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
LYRM4	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	66,3%	66,3%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chediak-Higashi syndrome, 214500
LYZ	SKIN DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Amyloidosis, renal, 105200
LZTFL1	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 17, 615994

LZTR1	HEART HEMOSTATIC/THROMBOTIC DISORDERS HNPD SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
LZTS1	MENDELIOME	100,0%	100,0%	Esophageal squamous cell carcinoma, somatic, 133239
M1AP	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 48, 619108
MAB21L1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	MENDELIOME	100,0%	100,0%	Prostate cancer, somatic, 176807 Lymphoma, somatic,
MAD2L2	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	?Fanconi anemia, complementation group V, 617243
MADD	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 DEEAH syndrome, 619004

MAF	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	94,5%	90,7%	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088
MAFA	MENDELIOME	100,0%	99,9%	Insulinomatosis and diabetes mellitus, 147630
MAFB	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	100,0%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	FETAL AKINESIA (AKI) INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Schaaf-Yang syndrome, 615547
MAGI2	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	98,4%	96,3%	Nephrotic syndrome, type 15, 617609
MAGT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	98,7%	98,7%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MAK	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 62, 614181
MAL2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MALT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 12, 615468

MAML2	MENDELIOME	100,0%	100,0%	Mucoepidermoid salivary gland carcinoma,
MAMLD1	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	Hypospadias 2, X-linked, 300758
MAN1B1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Rafiq syndrome, 614202
MAN2B1	HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MAN2C1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Congenital disorder of deglycosylation 2, 619775
MANBA	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mannosidosis, beta, 248510
MAOA	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,4%	Brunner syndrome, 300615
MAP1B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 83, 619808 Periventricular nodular heterotopia 9, 618918

MAP1LC3B2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MAP2K1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	46XY sex reversal 6, 613762
MAP3K14	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MAP3K20	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800
MAP3K8	MENDELIOME	100,0%	100,0%	Lung cancer, somatic, 211980
MAP4K4	MENDELIOME	100,0%	100,0%	No OMIM Disease ID

MAPK1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Noonan syndrome 13, 619087
MAPK8	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MAPK8IP3	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK3	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Macular dystrophy, patterned, 3, 617111
MAPKAPK5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MAPKBP1	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 20, 617271
MAPRE2	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	ALS MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MARCHF6	MENDELIOME	100,0%	100,0%	Epilepsy, familial adult myoclonic, 3, 613608
MARK3	MENDELIOME	100,0%	100,0%	?Visual impairment and progressive phthisis bulbi, 618283
MARS1	NEUROPATHIES LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Interstitial lung and liver disease, 615486 ?Trichothiodystrophy 9, nonphotosensitive, 619692 Charcot-Marie-Tooth disease, axonal, type 2U, 616280

MARS2	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 49, 610153
MASP1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	3MC syndrome 1, 257920
MASP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	MASP2 deficiency, 613791
MAST1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MASTL	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MAT1A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MAT2A	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MATN3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MATR3	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 21, 606070
MAX	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID

MBD5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 1, 156200
MBL2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MBOAT7	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MBTPS2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 IFAP syndrome with or without BRESHECK syndrome, 308205 ?Olmsted syndrome, X-linked, 300918
MC2R	DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	MENDELIOME	100,0%	100,0%	Obesity (BMIQ20), 618406
MCAT	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MCC	MENDELIOME	100,0%	100,0%	Colorectal cancer, somatic, 114500
MCCC1	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200

MCCC2	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Factor V and factor VIII, combined deficiency of, 613625
MCIDAS	CILIOPATHIES MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 42, 618695
MCM10	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 70, 616968
MCM3AP	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	95,5%	95,5%	Immunodeficiency 54, 609981
MCM5	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Meier-Gorlin syndrome 8, 617564
MCM6	MENDELIOME	100,0%	100,0%	Lactase persistence/nonpersistence, 223100

MCM8	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY HEREDITARY CANCER	94,4%	94,4%	?Premature ovarian failure 10, 612885
MCM9	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY HEREDITARY CANCER	100,0%	100,0%	Ovarian dysgenesis 4, 616185
MCOLN1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucolipidosis IV, 252650
MCPH1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 1, primary, autosomal recessive, 251200
MCTP2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MCUR1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
MDH1	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Developmental and epileptic encephalopathy 51, 617339
MDM2	MENDELIOME	92,6%	92,6%	?Lessel-Kubisch syndrome, 618681
MDM4	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	?Bone marrow failure syndrome 6, 618849

MECOM	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Encephalopathy, neonatal severe, 300673 Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	CRANIOFACIAL ANOMALIES SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Hardikar syndrome, 301068 Opitz-Kaveggia syndrome, 305450
MED12L	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Nizon-Isidor syndrome, 618872
MED13	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 61, 618009
MED13L	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MED17	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668

MED23	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MED27	INTELLECTUAL DISABILITY MENDELIOME	84,7%	84,7%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEF2C	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEFV	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	96,4%	96,4%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MEGF8	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Carpenter syndrome 2, 614976
MEI1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 22, 617706
MEIS2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Cleft palate, cardiac defects, and mental retardation, 600987

MEN1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Multiple endocrine neoplasia 1, 131100 Lipoma, somatic, Angiofibroma, somatic, Carcinoid tumor of lung, Adrenal adenoma, somatic, Parathyroid adenoma, somatic,
MEOX1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Klippel-Feil syndrome 2, 214300
MERTK	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,2%	99,1%	Retinitis pigmentosa 38, 613862
MESD	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XX, 618644
MESP2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	97,5%	97,5%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
EEF1AKNMT	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
METTL23	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,5%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFAP5	MENDELIOME	100,0%	100,0%	Aortic aneurysm, familial thoracic 9, 616166

MFF	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Keutel syndrome, 245150

MIA3	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MIB1	HEART MENDELIOME	100,0%	100,0%	Left ventricular noncompaction 7, 615092
MICOS13	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myopathy with extrapyramidal signs, 615673
MICU2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
MID1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Opitz GBBB syndrome, 300000
MID2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder, X-linked 101, 300928
MIEF2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 49, 619024
MINPP1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Pontocerebellar hypoplasia, type 16, 619527
MIP	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 15, multiple types, 615274
MIPEP	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 31, 617228

MIR140	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG	MENDELIOME	NC	NC	No OMIM Disease ID
MIR184	VISION DISORDERS MENDELIOME	NC	NC	EDICT syndrome, 614303
MIR204	VISION DISORDERS MENDELIOME	NC	NC	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96	HEARING IMPAIRMENT MENDELIOME	NC	NC	Deafness, autosomal dominant 50, 613074
MITF	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MELANOOM MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306
MKKS	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKRN3	DISORDERS OF SEX DEVELOPMENT MENDELIOME	96,0%	96,0%	Precocious puberty, central, 2, 615346

MKS1	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MLC1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MLH3	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MLIP	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	No OMIM Disease ID
MLLT10	MENDELIOME	97,1%	97,1%	Leukemia, acute myeloid, 601626
MLPH	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Griselli syndrome, type 3, 609227
MLYCD	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Malonyl-CoA decarboxylase deficiency, 248360

MMAA	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	89,7%	89,7%	Methylmalonic aciduria, cbID type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cbID type, 277410 Homocystinuria, cbID type, variant 1, 277410
MME	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	98,0%	98,0%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMGT1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MMP1	MENDELIOME	100,0%	100,0%	COPD, rate of decline of lung function in, 606963
MMP13	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	92,4%	92,4%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400

MMP14	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Winchester syndrome, 277950
MMP19	MENDELIOME	100,0%	100,0%	Cavitory optic disc anomalies, 611543
MMP2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Metaphyseal anadysplasia 2, 613073
MMUT	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria, mut(0) type, 251000
MN1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	CEBALID syndrome, 618774 Meningioma, 607174
MNS1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948

MNX1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	97,7%	91,6%	Currarino syndrome, 176450
MOCOS	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Xanthinuria, type II, 603592
MOCS1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Molybdenum cofactor deficiency A, 252150
MOCS2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Molybdenum cofactor deficiency B, 252160
MOG	MENDELIOME	100,0%	100,0%	?Narcolepsy 7, 614250
MOGS	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPC1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type If, 609180

MPDZ	HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPEG1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 77, 619223
MPI	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myeloperoxidase deficiency, 254600
MPV17	NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810

MPZ	FETAL AKINESIA (AKI) NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 2I, 607677 Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1B, 118200 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, type 2J, 607736
MPZL2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 111, 618145
MRAP	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glucocorticoid deficiency 2, 607398
MRAS	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Noonan syndrome 11, 618499
MRE11	MOVEMENT DISORDERS BREAST CANCER SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	98,9%	98,9%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
MRPL3	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

MRPL44	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
MRPS14	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 46, 618952
MRPS25	MENDELIOME MITOCHONDRIAL DISORDERS	82,7%	82,7%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	MENDELIOME MITOCHONDRIAL DISORDERS	86,6%	86,6%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
MRPS7	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

MRTFA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	92,8%	92,8%	?Immunodeficiency 66, 618847
MS4A1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Immunodeficiency, common variable, 5, 613495
MSH2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSH3	MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	99,1%	99,1%	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH4	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	No OMIM Disease ID
MSH5	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Premature ovarian failure 13, 617442
MSH6	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome 3, 619097
MSL2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MSL3	INTELLECTUAL DISABILITY MENDELIOME	98,4%	97,1%	Basilicata-Akhtar syndrome, 301032
MSMO1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, congenital cataract, and psoriasisiform dermatitis, 616834

MSN	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 50, 300988
MSR1	MENDELIOME	100,0%	100,0%	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 74, 613718
MSTN	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	?Muscle hypertrophy, 614160
MSTO1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, mitochondrial, and ataxia, 617675
MSX1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874
MSX2	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Parietal foramina with cleidocranial dysplasia, 168550 Craniosynostosis 2, 604757 Parietal foramina 1, 168500
MTAP	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780

MTHFR	MOVEMENT DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Homocystinuria due to MTHFR deficiency, 236250
MTHFS	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	FETAL AKINESIA (AKI) LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myopathy, centronuclear, X-linked, 310400
MTMR2	NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	94,3%	92,1%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTPAP	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940

C12orf65	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTRR	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Abetalipoproteinemia, 200100
MTX2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mandibuloacral dysplasia progeroid syndrome, 619127
MUC1	MENDELIOME	100,0%	100,0%	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000
MUC16	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MUSK	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659
MVD	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Porokeratosis 7, multiple types, 614714

MVK	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	90,5%	90,5%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MXI1	MENDELIOME	100,0%	100,0%	Prostate cancer, somatic, 176807 Neurofibrosarcoma, somatic,
MYBPC1	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915 Arthrogryposis, distal, type 1B, 614335
MYBPC3	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MYC	MENDELIOME	100,0%	100,0%	Burkitt lymphoma, somatic, 113970
MYCN	FETAL AKINESIA (AKI) SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Feingold syndrome 1, 164280
MYD88	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYF5	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH11	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350

MYH14	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME	100,0%	100,0%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	FETAL AKINESIA (AKI) SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1A, 178110 Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYH6	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251
MYH7	CONGENITAL HEART DISEASE HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Laing distal myopathy, 160500 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160
MYH7B	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MYH8	FETAL AKINESIA (AKI) SKIN DISORDERS MENDELIOME	100,0%	100,0%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300

MYH9	HEREDITARY BONE MARROW FAILURE HEARING IMPAIRMENT HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYL1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYLPF	FETAL AKINESIA (AKI) SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Arthrogryposis, distal, type 1C, 619110
MYL2	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,0%	Cardiomyopathy, hypertrophic, 10, 608758 Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	HEART MENDELIOME	100,0%	100,0%	?Atrial fibrillation, familial, 18, 617280
MYL7	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MYL9	MENDELIOME	100,0%	100,0%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365
MYLK	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 Aortic aneurysm, familial thoracic 7, 613780
MYLK2	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID

MYMK	MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Carey-Fineman-Ziter syndrome, 254940
MYO15A	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 3, 600316
MYO18B	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1A	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MYO1E	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glomerulosclerosis, focal segmental, 6, 614131
MYO1H	MENDELIOME	100,0%	100,0%	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482
MYO3A	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 30, 607101
MYO5A	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Griselli syndrome, type 1, 214450
MYO5B	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diarrhea 2, with microvillus atrophy, 251850
MYO6	HEARING IMPAIRMENT HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900 Deafness, autosomal dominant 11, 601317

MYO9A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	VISION DISORDERS MENDELIOME	100,0%	100,0%	Glaucoma 1A, primary open angle, 137750
MYOCD	MENDELIOME	100,0%	100,0%	Megabladder, congenital, 618719
MYOD1	MENDELIOME	100,0%	100,0%	Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies, 618975
MYOF	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Angioedema, hereditary, 7, 619366
MYOM1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
MYORG	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
MYOT	HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	CONGENITAL HEART DISEASE DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME	100,0%	100,0%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280

MYSM1	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	96,4%	96,4%	Bone marrow failure syndrome 4, 618116
MYT1L	INTELLECTUAL DISABILITY MENDELIOME	90,2%	90,2%	Intellectual developmental disorder, autosomal dominant 39, 616521
NAA10	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME	96,8%	96,8%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 73, 619717
NACC1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGA	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241

NAGLU	NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	N-acetylglutamate synthase deficiency, 237310
NALCN	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,8%	99,8%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	MENDELIOME	100,0%	99,9%	Spermatogenic failure 12, 615413
NANS	MOVEMENT DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NARS2	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAT8L	MENDELIOME PRE CONCEPTION SCREENING	99,8%	98,7%	?N-acetylaspartate deficiency, 614063

NAXD	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NBEA	EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gray platelet syndrome, 139090
NBN	HEREDITARY BONE MARROW FAILURE BREAST CANCER PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260

NCAPD2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Khan-Khan-Katsanis syndrome, 618460
NCAPH	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 23, primary, autosomal recessive, 617985
NCDN	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCF1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NCKAP1L	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 72 with autoinflammation, 618982
NCOA3	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NCOA4	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID

NCSTN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Acne inversa, familial, 1, 142690
NDE1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	MENDELIOME	100,0%	100,0%	Prader-Willi syndrome, 176270
NDNF	MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NDP	VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 23, 618244

NDUFA13	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	MENDELIOME MITOCHONDRIAL DISORDERS	92,2%	88,6%	No OMIM Disease ID
NDUFA4	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFA6	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFA8	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFAF1	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 10, 618233

NDUFAF3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 17, 618239 Fanconi renotubular syndrome 5, 618913
NDUFAF7	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFAF8	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFB10	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	VISION DISORDERS SKIN DISORDERS HEART IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	99,9%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFB3	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 25, 618246

NDUFB4	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFB5	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFB6	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFB7	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFB8	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	98,7%	98,7%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFC2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	95,3%	91,3%	Mitochondrial complex I deficiency, nuclear type 8, 618230

NDUFS4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NDUFS6	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NEB	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	99,9%	99,9%	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334

NEBL	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NECAP1	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Periventricular nodular heterotopia 7, 617201
NEFH	NEUROPATHIES MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	ALS VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK10	MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 44, 618781

NEK11	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NEK2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,1%	96,1%	?Retinitis pigmentosa 67, 615565
NEK8	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824
NEK9	FETAL AKINESIA (AKI) SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NEMF	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEPRO	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anauxetic dysplasia 3, 618853
NEU1	MOVEMENT DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROD1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Maturity-onset diabetes of the young 6, 606394
NEUROD2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 72, 618374
NEUROG3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diarrhea 4, malabsorptive, congenital, 610370

NEXMIF	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 98, 300912
NEXN	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NF2	MOVEMENT DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091
NFASC	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFAT5	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NFATC1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NFE2	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NFE2L2	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744

NFIA	INTELLECTUAL DISABILITY MENDELIOME	99,2%	99,2%	Brain malformations with or without urinary tract defects, 613735
NFIB	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,7%	Marshall-Smith syndrome, 602535 Malan syndrome, 614753
NFKB1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency, common variable, 12, 616576
NFKB2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency, common variable, 10, 615577
NFKBIA	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Ectodermal dysplasia and immunodeficiency 2, 612132
NFS1	MENDELIOME MITOCHONDRIAL DISORDERS	89,5%	89,5%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of deglycosylation 1, 615273

NHEJ1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 6112
NHLH2	MENDELIOME	100,0%	100,0%	?Hypogonadotropic hypogonadism 27 without anosmia, 619755
NHLRC1	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	FINCA syndrome, 618278
NHP2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	99,1%	99,1%	?Seckel syndrome 7, 614851
NIPA1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 6, 612281

NIPBL	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Cornelia de Lange syndrome 1, 122470
NKAP	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
NKX2-1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Hypoplastic left heart syndrome 2, 614435 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Conotruncal heart malformations, variable, 217095 Ventricular septal defect 3, 614432 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	CONGENITAL HEART DISEASE HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NLGN3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

NLGN4X	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, 300495
NLRP4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	CINCA syndrome, 607115 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Deafness, autosomal dominant 34, with or without inflammation, 617772 Muckle-Wells syndrome, 191900
NLRP6	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NLRP7	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hydatidiform mole, recurrent, 1, 231090
NME1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NME3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
NME5	CILIOPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NME8	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 6, 610852

NMNAT1	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	98,5%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NMNAT2	NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NNT	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,4%	96,4%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Premature ovarian failure 5, 611548
NOD2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Blau syndrome, 186580
NODAL	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME	100,0%	100,0%	Heterotaxy, visceral, 5, 270100
NOG	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Symphalangism, proximal, 1A, 185800 Brachydactyly, type B2, 611377 Stapes ankylosis with broad thumbs and toes, 184460 Tarsal-carpal coalition syndrome, 186570 Multiple synostoses syndrome 1, 186500
NOL3	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	?Myoclonus, familial, 1, 614937
NONO	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 34, 300967

NOP10	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 36, 614153
NOS1AP	HEART RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephrotic syndrome, type 22, 619155
NOS2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NOTCH1	CONGENITAL HEART DISEASE SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH2NLC	MENDELIOME	100,0%	100,0%	Tremor, hereditary essential, 6, 618866 Oculopharyngodistal myopathy 3, 619473 Neuronal intranuclear inclusion disease, 603472

NOTCH3	MENDELIOME	100,0%	100,0%	Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310
NOVA2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPAT	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NPC1	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Niemann-pick disease, type C2, 607625
NPHP1	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	VISION DISORDERS CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966

NPHS1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 1, 256300
NPHS2	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 2, 600995
NPL	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NPM1	HEREDITARY BONE MARROW FAILURE DKC MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Leukemia, acute myeloid, somatic, 601626
NPPA	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPPB	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NPPC	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NPR2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255 Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Boudin-Mortier syndrome, 619543
NPRL2	EPILEPSY MENDELIOME	100,0%	100,0%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	EPILEPSY MENDELIOME	100,0%	100,0%	Epilepsy, familial focal, with variable foci 3, 617118

NR0B1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MALE INFERTILITY MENDELIOME	100,0%	100,0%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR0B2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Obesity, mild, early-onset, 601665
NR1H4	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 37, 611131 Enhanced S-cone syndrome, 268100
NR2F1	VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	99,8%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779
NR3C1	DISORDERS OF SEX DEVELOPMENT MENDELIOME	100,0%	100,0%	Glucocorticoid resistance, 615962
NR3C2	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME	100,0%	100,0%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NR4A3	MENDELIOME	100,0%	100,0%	Chondrosarcoma, extraskeletal myxoid, 612237

NR5A1	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Spermatogenic failure 8, 613957
NRAP	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NRAS	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
NRCAM	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NRIP1	MENDELIOME	100,0%	100,0%	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type,
NRROS	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pitt-Hopkins-like syndrome 2, 614325

NSD1	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Sotos syndrome, 117550
NSD2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Rauch-Steindl syndrome, 619695
NSDHL	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	CK syndrome, 300831 CHILD syndrome, 308050
NSF	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 96, 619340
NSMCE2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Seckel syndrome 10, 617253
NSMCE3	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 5, 611091

NSUN3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 48, 619012
NT5C2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Calcification of joints and arteries, 211800
NTF4	MENDELIOME	100,0%	100,0%	Glaucoma 1, open angle, 1O, 613100
NTHL1	MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Familial adenomatous polyposis 3, 616415
NTN1	MENDELIOME	100,0%	100,0%	Mirror movements 4, 618264
NTNG2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUAK2	MENDELIOME	100,0%	100,0%	?Anencephaly 2, 619452
NUBPL	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 21, 618242

NUDT2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NUMA1	MENDELIOME	100,0%	100,0%	Leukemia, acute promyelocytic, somatic, 612376
NUP107	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP133	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177
NUP155	HEART MENDELIOME	100,0%	100,0%	?Atrial fibrillation 15, 615770
NUP160	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Nephrotic syndrome, type 19, 618178
NUP188	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sandestig-Stefanova syndrome, 618804
NUP205	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Nephrotic syndrome, type 13, 616893
NUP214	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukemia, T-cell acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, somatic, 601626
NUP37	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Striatonigral degeneration, infantile, 271930
NUP85	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 17, 618176

NUP88	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fetal akinesia deformation sequence 4, 618393
NUP93	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	95,5%	95,5%	Nephrotic syndrome, type 12, 616892
NUS1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
NUTM2B-AS1	MENDELIOME	NC	NC	?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXF5	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
NXN	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Robinow syndrome, autosomal recessive 2, 618529
NYX	VISION DISORDERS MENDELIOME	100,0%	100,0%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAS1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
OAT	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSCN	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID

OBSL1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	3-M syndrome 2, 612921
OCA2	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200
OCLN	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudo-TORCH syndrome 1, 251290
OCRL	VISION DISORDERS CILIOPATHIES HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Dent disease 2, 300555 Lowe syndrome, 309000
CCDC114	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 20, 615067
ARMC4	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	96,3%	96,3%	Ciliary dyskinesia, primary, 23, 615451
CCDC151	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 30, 616037
TTC25	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 35, 617092
ODAM	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID

ODAPH	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IIA4, 614832
ODC1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Bachmann-Bupp syndrome, 619075
OFD1	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OGDH	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Yoon-Bellen neurodevelopmental syndrome, 619701
OGG1	MENDELIOME	100,0%	100,0%	Renal cell carcinoma, clear cell, somatic, 144700
OGT	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 106, 300997
OPA1	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896

OPA3	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPCML	MENDELIOME	100,0%	100,0%	Ovarian cancer, somatic, 167000
OPHN1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,5%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
OPLAH	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	5-oxoprolinase deficiency, 260005
OPN1LW	VISION DISORDERS MENDELIOME	96,2%	95,8%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	VISION DISORDERS MENDELIOME	99,1%	98,7%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPN1SW	MENDELIOME	100,0%	100,0%	Colorblindness, tritan, 190900
OPTN	ALS VISION DISORDERS MENDELIOME	100,0%	100,0%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435
ORAI1	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883

ORC1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Meier-Gorlin syndrome 1, 224690
ORC4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Meier-Gorlin syndrome 2, 613800
ORC6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Meier-Gorlin syndrome 3, 613803
OSBPL2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 67, 616340
OSGEP	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galloway-Mowat syndrome 3, 617729
OSMR	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteopetrosis, autosomal recessive 5, 259720
OTC	METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME	100,0%	100,0%	Ornithine transcarbamylase deficiency, 311250
OTOA	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 22, 607039

OTOF	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 18B, 614945
OTOGL	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 84B, 614944
OTUD5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,7%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	INTELLECTUAL DISABILITY MENDELIOME	99,7%	98,7%	No OMIM Disease ID
OTULIN	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OTX2	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125
OVOL2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXA1L	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
OXCT1	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050

OXR1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P2RX2	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 41, 608224
P2RY12	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bleeding disorder, platelet-type, 8, 609821
P3H1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type VIII, 610915
P3H2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Myopia 25, autosomal dominant, 617238
P4HB	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Cole-Carpenter syndrome 1, 112240
P4HTM	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 6184
PABPN1	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Oculopharyngeal muscular dystrophy, 164300
PACS1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Schuurs-Hoeijmakers syndrome, 615009

PACS2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 66, 618067
PADI3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Uncombable hair syndrome, 191480
PADI6	MENDELIOME	100,0%	100,0%	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Phenylketonuria, 261600
PAK1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 30, 300558
PALB2	HEREDITARY BONE MARROW FAILURE BREAST CANCER SKIN DISORDERS HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group N, 610832

MPP5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PAM16	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	82,9%	82,9%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	MOVEMENT DISORDERS VISION DISORDERS IRON DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANK4	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Cataract 49, 619593
PANX1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Oocyte maturation defect 7, 618550
PAPPA2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Short stature, Dauber-Argente type, 619489
PAPSS2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinson disease 7, autosomal recessive early-onset, 606324

PARN	HEREDITARY BONE MARROW FAILURE DKC PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	89,5%	87,8%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARP4	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PARP6	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PARS2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 75, 618437
PATL2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Oocyte maturation defect 4, 617743
PAX1	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Otofaciocervical syndrome 2, 615560
PAX2	VISION DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX3	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220

PAX4	MENDELIOME	100,0%	100,0%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853
PAX5	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
PAX6	MOVEMENT DISORDERS VISION DISORDERS CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200 ?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190
PAX7	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Rhabdomyosarcoma 2, alveolar, 268220 Myopathy, congenital, progressive, with scoliosis, 618578
PAX8	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	100,0%	100,0%	Tooth agenesis, selective, 3, 604625
PBRM1	MENDELIOME	100,0%	100,0%	?Renal cell carcinoma, clear cell, 144700
PBX1	DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or development of the central nervous system, 607000

PC	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pyruvate carboxylase deficiency, 266150
PCARE	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 54, 613428
PCBD1	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	HEART PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Propionicacidemia, 606054
PCCB	HEART PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	99,9%	98,1%	Propionicacidemia, 606054
PCDH12	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1F, 602083

PCDH19	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PCGF2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Turnpenny-Fry syndrome, 618371
PCK1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
PCLO	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Obesity with impaired prohormone processing, 600955
PCSK9	MENDELIOME	100,0%	100,0%	Hypercholesterolemia, familial, 3, 603776

PCYT1A	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PCYT2	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spastic paraplegia 82, autosomal recessive, 618770
PDCD1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PDCD10	MENDELIOME	100,0%	100,0%	Cerebral cavernous malformations-3, 603285
PDE10A	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	90,0%	87,2%	Striatal degeneration, autosomal dominant, 616922 Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE11A	MENDELIOME	100,0%	100,0%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 74, 618140
PDE2A	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE3A	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Hypertension and brachydactyly syndrome, 112410
PDE4D	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 43, 613810

PDE6B	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa-40, 613801 Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone dystrophy 4, 613093
PDE6D	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 22, 615665
PDE6G	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 57, 613582
PDE6H	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDE8B	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	MOVEMENT DISORDERS SKIN DISORDERS MENDELIOME PARKINSON HEREDITARY CANCER	100,0%	100,0%	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907
PDGFRA	VISION DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	MOVEMENT DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON	100,0%	100,0%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007
PDGFRL	MENDELIOME	100,0%	100,0%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500

PDHA1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
PDK2	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
PDK3	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
PDLIM3	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PDLIM5	HEART MENDELIOME	98,7%	96,1%	No OMIM Disease ID
PDP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pyruvate dehydrogenase phosphatase deficiency, 608782

PDSS1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	97,4%	97,4%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 23, 610245
PDZD7	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PEPD	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Prolidase deficiency, 170100
PER2	MENDELIOME	100,0%	100,0%	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	MENDELIOME	100,0%	100,0%	?Advanced sleep phase syndrome, familial, 3, 616882
PERCC1	MENDELIOME	100,0%	100,0%	Diarrhea 11, malabsorptive, congenital, 618662

PERP	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Erythrokeratoderma variabilis et progressiva 7, 619209 Olmsted syndrome 2, 619208
PET100	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PEX1	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 14B, 614920
PEX12	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859

PEX13	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	EPILEPSY NEUROPATHIES PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	MOVEMENT DISORDERS VISION DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867

PEX26	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	EPILEPSY HEART SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617

PEX7	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS HEART NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease VII, 232800
PFN1	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Glycogen storage disease X, 261670
PGAP1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 4, 615716

PGK1	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	CRANIOFACIAL ANOMALIES HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS	94,2%	94,2%	Congenital disorder of glycosylation, type Ia, 614921
PGM2L1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PGM3	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	91,7%	91,7%	Immunodeficiency 23, 615816
PHACTR1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 70, 618298
PHC1	MENDELIOME	100,0%	100,0%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	100,0%	99,9%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF21A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without sei

PHF6	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263
PHGDH	FETAL AKINESIA (AKI) SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	Chung-Jansen syndrome, 617991
PHKA1	HEART METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	99,9%	Muscle glycogenosis, 300559
PHKA2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PHKG2	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycogen storage disease IXc, 613027

PHOX2A	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880
PHYH	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS HEART NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Refsum disease, 266500
PI4K2A	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PI4KA	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Spastic paraplegia 84, autosomal recessive, 619621 Gastrointestinal defects and immunodeficiency syndrome 2, 619708 Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PI4KB	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PIBF1	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 33, 617767
PICALM	MENDELIOME	100,0%	100,0%	Leukemia, acute myeloid, somatic, 601626
PIDD1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PIEZ01	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380

PIEZ02	FETAL AKINESIA (AKI) NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS	100,0%	100,0%	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIGA	SKIN DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 80, 618580
PIGC	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356
PIGG	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	80,3%	74,7%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879

PIGL	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	CHIME syndrome, 280000
PIGM	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	98,8%	98,8%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 95, 618143
PIGT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590

PIGV	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3C2A	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Oculoskeletal dental syndrome, 618440
PIK3CA	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Non-small cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108 Macrodactyly, somatic,,
PIK3CD	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 97 with autoinflammation, 619802

PIK3R1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	VISION DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Corneal fleck dystrophy, 121850
PINK1	MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinson disease 6, early onset, 605909
PIP5K1C	FETAL AKINESIA (AKI) METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Lethal congenital contractual syndrome 3, 611369
PISD	HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Liberfarb syndrome, 618889
PITPNM3	MENDELIOME	100,0%	100,0%	Cone-rod dystrophy 5, 600977
PITRM1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 30, 619405

PITX1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PITX3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 11, multiple types, 610623 Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623
PIWIL2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PJA1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PJVK	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 59, 610220
PKD1	CILIOPATHIES LIVER DISORDERS MALE INFERTILITY RENAL DISORDERS MENDELIOME	99,9%	99,7%	Polycystic kidney disease 1, 173900
PKD1L1	CONGENITAL HEART DISEASE HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Polycystic kidney disease 2, 613095

PKDCC	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Rhizomelic limb shortening with dysmorphic features, 618821
PKHD1	CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	HEART MENDELIOME	95,0%	95,0%	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLA2G4A	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G5	VISION DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLA2G6	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600

PLA2G7	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Adenomas, salivary gland pleomorphic, somatic, 181030 Silver-Russell syndrome 4, 618907
PLAT	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLAU	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Quebec platelet disorder, 601709
PLCB1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 12, 613722
PLCB3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Auriculocondylar syndrome 2, 614669

PLCD1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 3, 610725
PLCG2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLCZ1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 17, 617214
PLD1	CONGENITAL HEART DISEASE HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cardiac valvular defect, developmental, 212093
PLD3	NEUROPATHIES MENDELIOME	100,0%	100,0%	?Spinocerebellar ataxia 46, 617770
PLEC	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 Epidermolysis bullosa simplex 5A, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	96,3%	96,3%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107

PLEKHM2	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLG	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PLIN1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Lipodystrophy, familial partial, type 4, 613877
PLK1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLK4	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MALE INFERTILITY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	FETAL AKINESIA (AKI) SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bruck syndrome 2, 609220

PLOD3	SKIN DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLS1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 76, 618787
PLS3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	97,2%	97,2%	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diarrhea 10, protein-losing enteropathy type, 618183
PLXNA1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLXNA2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PLXND1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

PMEPA1	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PMFBP1	CILIOPATHIES MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 31, 618112
PML	MENDELIOME	100,0%	100,0%	Leukemia, acute promyelocytic, PML/RARA type,
PMM2	MOVEMENT DISORDERS EPILEPSY HEART NEUROPATHIES PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	NEUROPATHIES MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	MOVEMENT DISORDERS NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, type 1A, 118220 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 1E, 118300 ?Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Dejerine-Sottas disease, 145900
PMPCA	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 6, 617954

PMS2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PMS2CL	MENDELIOME HEREDITARY CANCER	NC	NC	No OMIM Disease ID
PMVK	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Porokeratosis 1, multiple types, 175800
PNKD	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLDC1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 57, 619528
PNLIP	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Pancreatic lipase deficiency, 614338
PNMT	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PNP	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179

PNPLA1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	SKIN DISORDERS HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	MOVEMENT DISORDERS VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
PNPLA8	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POC1A	CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 20, 615973

POC5	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PODXL	RENAL DISORDERS MENDELIOME	94,3%	94,3%	No OMIM Disease ID
POF1B	MENDELIOME	100,0%	100,0%	?Premature ovarian failure 2B, 300604
POFUT1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Dowling-Degos disease 2, 615327
POGLUT1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POGZ	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	White-Sutton syndrome, 616364
POLA1	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLE2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

POLG	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY HYPOGONADOTROPIC HYPOGONADISM NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY MUSCLE DISORDERS	100,0%	100,0%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	100,0%	100,0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POLH	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, variant type, 278750
POLL	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
POLR1A	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1B	MENDELIOME	100,0%	100,0%	Treacher-Collins syndrome 4, 618939

POLR1C	MOVEMENT DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	83,0%	82,8%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Treacher Collins syndrome 2, 613717
POLR2A	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	MOVEMENT DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742

POLR3GL	MENDELIOME	100,0%	100,0%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POLR3K	MENDELIOME	100,0%	100,0%	Leukodystrophy, hypomyelinating, 21, 619310
POLRMT	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 55, 619743
POMC	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155

POMT2	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
POP1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anauxetic dysplasia 2, 617396
POPC3	MENDELIOME	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848
POR	CRANIOFACIAL ANOMALIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Focal dermal hypoplasia, 305600
POT1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES MELANO瘤 MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
POU1F1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU2AF1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

POU3F3	INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,0%	Snijders Blok-Fisher syndrome, 618604
POU3F4	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, X-linked 2, 304400
POU4F1	MENDELIOME	94,9%	91,9%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352
POU4F3	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 15, 602459
POU6F2	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
PPA2	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	METABOLIC DISORDERS MENDELIOME	98,3%	98,3%	Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 Carotid intimal medial thickness 1, 609338
PPCS	HEART METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Cardiomyopathy, dilated, 2C, 618189
PPIB	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type IX, 259440
PPIL1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 14, 619301
PPIP5K2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 100, 618422

PPM1D	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPM1K	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Maple syrup urine disease, mild variant, 615135
PPOX	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Porphyria variegata, 176200
PPP1CB	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PPP1R3A	MENDELIOME	100,0%	100,0%	Insulin resistance, severe, digenic, 125853
PPP2CA	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	93,6%	93,6%	Intellectual developmental disorder, autosomal dominant 36, 616362

PPP2R1B	MENDELIOME	100,0%	100,0%	Lung cancer, somatic, 211980
PPP2R2B	MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 12, 604326
PPP2R3C	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 Spermatogenic failure 36, 618420
PPP2R5B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PPP2R5C	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PPP2R5D	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 35, 616355
PPP3CA	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Developmental and epileptic encephalopathy 91, 617711
PPT1	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	82,5%	82,5%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Renpenning syndrome, 309500
PRCC	MENDELIOME	100,0%	100,0%	Renal cell carcinoma, papillary, 605074
PRCD	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 36, 610599

PRDM12	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	96,2%	94,0%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM15	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	No OMIM Disease ID
PRDM16	HEART MENDELIOME	100,0%	100,0%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Brittle cornea syndrome 2, 614170
PRDM6	HEART MENDELIOME	100,0%	100,0%	Patent ductus arteriosus 3, 617039
PRDM8	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cbIC type, digenic, 277400
PRDX2	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PRDX3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
PREPL	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 22, 616224

PRF1	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE EPILEPSY PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRG4	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PRIMPOL	VISION DISORDERS MENDELIOME	100,0%	100,0%	Myopia 22, autosomal dominant, 615420
PRKAA1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
PRKACA	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 Cardioacrofacial dysplasia 1, 619142
PRKACB	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cardioacrofacial dysplasia 2, 619143
PRKACG	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	HEART METABOLIC DISORDERS MENDELIOME	100,0%	99,9%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858

PRKAR1A	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,
PRKAR1B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Marbach-Schaaf neurodevelopmental syndrome, 619680
PRKCA	MENDELIOME	100,0%	100,0%	Pituitary tumor, invasive,
PRKCB	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PRKCD	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 14, 605361
PRKCSH	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Polycystic liver disease 1, 174050
PRKD1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	MENDELIOME	92,7%	92,7%	Aortic aneurysm, familial thoracic 8, 615436

PRKG2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638 Acromesomelic dysplasia 4, 619636
PRKN	MENDELIOME PARKINSON PRE CONCEPTION SCREENING HEREDITARY CANCER	75,5%	75,3%	Adenocarcinoma of lung, somatic, 211980 Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000
PRKRA	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Dystonia 16, 612067
PRLR	MENDELIOME	100,0%	100,0%	Multiple fibroadenomas of the breast, 615554 Hyperprolactinemia, 615555
PRMT7	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	NEUROPATHIES MENDELIOME	100,0%	100,0%	Spongiform encephalopathy with neuropsychiatric features, 606688 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400
PROC	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304
PRODH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperprolinemia, type I, 239500
PROK2	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628

PROKR2	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657
PROP1	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pituitary hormone deficiency, combined, 2, 262600
PRORP	HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 54, 619737
PROS1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	98,4%	98,4%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336
PROZ	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PRPF3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 18, 601414
PRPF31	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 11, 600138
PRPF4	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 70, 615922

PRPF6	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 60, 613983
PRPF8	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 13, 600059
PRPH2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Macular dystrophy, patterned, 1, 169150 Choroidal dystrophy, central areolar 2, 613105 Retinitis punctata albescens, 136880 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133
PRPS1	HEARING IMPAIRMENT NEUROPATHIES PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	100,0%	100,0%	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PRR11	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PRR12	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neuroocular syndrome, 619539
PRRT2	MOVEMENT DISORDERS EPILEPSY MENDELIOME	100,0%	100,0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Seizures, benign familial infantile, 2, 605751 Episodic kinesigenic dyskinesia 1, 128200
PRRX1	CRANIOFACIAL ANOMALIES MENDELIOME SCHISIS	100,0%	100,0%	Agnathia-otocephaly complex, 202650
PRSS1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Pancreatitis, hereditary, 167800
PRSS12	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 1, 249500

PRSS56	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, isolated 6, 613517
PRUNE1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	93,6%	93,6%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	97,9%	96,8%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PSAT1	FETAL AKINESIA (AKI) SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSEN1	SKIN DISORDERS MENDELIOME PARKINSON	100,0%	100,0%	Pick disease, 172700 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 ?Acne inversa, familial, 3, 613737 Cardiomyopathy, dilated, 1U, 613694 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, 607822
PSEN2	MENDELIOME	100,0%	100,0%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSIP1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID

PSMA3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PSMB1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PSMB10	MENDELIOME	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 5, 619175
PSMB4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3	MENDELIOME	100,0%	100,0%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354
PSMC3IP	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Ovarian dysgenesis 3, 614324
PSMC5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PSMD12	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Stankiewicz-Isidor syndrome, 617516
PSMG2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSPH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Phosphoserine phosphatase deficiency, 614023

PSTPIP1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCD3	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 51, 619057
PTCH1	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTCH2	SKIN DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Medulloblastoma, somatic, 155255 Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462
PTCHD1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PTDSS1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	BREAST CANCER SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Cowden syndrome 1, 158350 Lhermitte-Duclos disease, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTF1A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTGIS	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Hypertension, essential, 145500

PTGS1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PTH	MENDELIOME	100,0%	100,0%	Hypoparathyroidism, familial isolated 1, 146200
PTH1R	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTHLH	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Brachydactyly, type E2, 613382
PTPN11	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	MENDELIOME	100,0%	100,0%	Colon cancer, somatic, 114500
PTPN14	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Choanal atresia and lymphedema, 613611

PTPN22	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
PTPN23	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPRC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRF	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Colon cancer, somatic, 114500
PTPRO	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 6, 614196
PTPRQ	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	92,8%	92,6%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	FETAL AKINESIA (AKI) MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

PTS	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Verheij syndrome, 615583
PUM1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 47, 617931
PURA	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	IRON DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	99,2%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUSS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anterior segment dysgenesis 7, with sclerocornea, 269400

PYCR1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycogen storage disease VI, 232700
PYGM	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	McArdle disease, 232600
PYROXD1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Myopathy, myofibrillar, 8, 617258
QARS1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, C, 261630

QRICH1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Ververi-Brady syndrome, 617982
QRICH2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 35, 618341
QSLR1	HEART MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Combined oxidative phosphorylation deficiency 40, 618835
RAB11B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RAB18	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Warburg micro syndrome 3, 614222
RAB23	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Carpenter syndrome, 201000
RAB27A	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Griselli syndrome, type 2, 607624
RAB28	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 18, 615374

RAB33B	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Smith-McCort dysplasia 2, 615222
RAB39B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 72, 300271 Waisman syndrome, 311510
RAB3GAP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,4%	99,4%	Martsolf syndrome 2, 619420 Warburg micro syndrome 1, 600118
RAB3GAP2	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225
RAB7A	NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAC2	PRIMARY IMMUNODEFICIENCIES MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 73A with defective neutrophil chemotaxis and leukocytosis, 608203 ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAC3	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RACGAP1	MENDELIOME	100,0%	100,0%	?Anemia, congenital dyserythropoietic, type IIb, autosomal recessive, 619789

RAD21	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RAD21L1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RAD50	BREAST CANCER SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE MENDELIOME	89,4%	89,4%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51C	HEREDITARY BONE MARROW FAILURE BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group O, 613390
RAD51D	BREAST CANCER HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
RAD54B	MENDELIOME	100,0%	100,0%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	MENDELIOME	100,0%	100,0%	Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic,

RAF1	CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAG1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
RAG2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Smith-Magenis syndrome, 182290
RALA	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermoregulation, 618797
RANBP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID

RANGRF	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RAP1GDS1	MENDELIOME	100,0%	100,0%	Lymphocytic leukemia, acute T-cell,
RAPGEF2	MENDELIOME	100,0%	100,0%	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, syndromic 12, 615524
RARS1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	94,4%	94,4%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	MENDELIOME	100,0%	100,0%	Capillary malformation-arteriovenous malformation 1, 608354 Basal cell carcinoma, somatic, 605462
RASGRP1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 64, 618534
RASGRP2	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Bleeding disorder, platelet-type, 18, 615888

RAX	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, isolated 3, 611038
RAX2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757
RB1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RB1CC1	MENDELIOME	100,0%	100,0%	Breast cancer, somatic, 114480
RBBP6	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RBBP8	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBCK1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,7%	No OMIM Disease ID
RBL2	MENDELIOME	100,0%	100,0%	Brunet-Wagner neurodevelopmental syndrome, 619690
RBM10	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	TARP syndrome, 311900

RBM20	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder, X-linked, syndromic 11, Shashi type, 300238
RBP3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Retinitis pigmentosa 66, 615233
RBP4	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Adams-Oliver syndrome 3, 614814
RC3H1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RCBTB1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinal dystrophy with or without extraocular anomalies, 617175

RD3	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 12, 610612
RDH11	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 13, 612712
RDH5	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fundus albipunctatus, 136880
RDX	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 24, 611022
REC114	MENDELIOME	100,0%	100,0%	Oocyte maturation defect 10, 619176
REC8	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	No OMIM Disease ID
RECQL4	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REEP1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
REEP2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625

REEP6	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,1%	94,0%	Retinitis pigmentosa 77, 617304
REL	PRIMARY IMMUNODEFICIENCIES MENDELIOME	99,9%	98,3%	Immunodeficiency 92, 619652
RELA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Mucocutaneous ulceration, chronic, 618287
RELB	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Immunodeficiency 53, 617585
RELN	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lissencephaly 2 (Norman-Roberts type), 257320
RELT	MENDELIOME	100,0%	100,0%	Amelogenesis imperfecta, type IIIC, 618386
REN	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
REPS1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,9%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	HEARING IMPAIRMENT MENDELIOME HEREDITARY CANCER	98,6%	98,6%	?Deafness, autosomal dominant 27, 612431 Fibromatosis, gingival, 5, 617626
RET	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIB, 162300
RETREG1	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
REV3L	INTELLECTUAL DISABILITY MENDELIOME	97,8%	97,6%	No OMIM Disease ID

RFC1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type I <sub>n</sub> , 612015
RFWD3	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Fanconi anemia, complementation group W, 617784
RFX3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RFX4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RFX5	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mitchell-Riley syndrome, 615710
RFX7	INTELLECTUAL DISABILITY MENDELIOME	99,1%	99,1%	No OMIM Disease ID
RFXANK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	MHC class II deficiency, complementation group B, 209920
RFXAP	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	MENDELIOME PRE CONCEPTION SCREENING	99,1%	99,0%	Retinitis pigmentosa 44, 613769

RGS10	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RGS9	VISION DISORDERS MENDELIOME	100,0%	100,0%	Bradyopsia, 608415
RGS9BP	VISION DISORDERS MENDELIOME	100,0%	100,0%	Bradyopsia, 608415
RHAG	MENDELIOME	100,0%	100,0%	Overhydrated hereditary stomatocytosis, 185000 Anemia, hemolytic, Rh-null, regulator type, 268150
RHBDF2	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Tylosis with esophageal cancer, 148500
RHCE	MENDELIOME	97,1%	97,0%	Rh-null disease, amorph type, 617970
RHEB	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RHO	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RHOA	SKIN DISORDERS MENDELIOME	80,7%	80,7%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RHOBTB2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 64, 618004
RHOG	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RHOH	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RIC1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	CATIFA syndrome, 618761
RILPL1	MENDELIOME	100,0%	100,0%	Oculopharyngodistal myopathy 4, 619790

RIMS1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cone-rod dystrophy 7, 603649
RIMS2	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	97,8%	97,8%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Infantile liver failure syndrome 3, 618641
RIPK1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	FETAL AKINESIA (AKI) CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIPOR2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 21, 607017 ?Deafness, autosomal recessive 104, 616515
RIPPLY2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spondylocostal dysostosis 6, 616566

RIT1	HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome 8, 615355
RLBP1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880 Fundus albipunctatus, 136880
RLIM	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Tonne-Kalscheuer syndrome, 300978
RMND1	HEARING IMPAIRMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID) HEREDITARY CANCER	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479

RNASEH2A	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	91,0%	91,0%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Prostate cancer 1, 601518
RNASET2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Tenorio syndrome, 616260
RNF13	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 73, 618379
RNF139	MENDELIOME	100,0%	100,0%	Renal cell carcinoma, 144700

RNF168	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	RIDDLE syndrome, 611943
RNF170	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	Ataxia, sensory, 1, autosomal dominant, 608984 Spastic paraplegia 85, autosomal recessive, 619686
RNF2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Luo-Schoch-Yamamoto syndrome, 619460
RNF212	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 62, 619673 Recombination rate QTL 1, 612042
RNF216	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF220	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNF31	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RNF43	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	MENDELIOME	100,0%	100,0%	Esophageal carcinoma, somatic, 133239
RNPC3	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pituitary hormone deficiency, combined or isolated, 7, 618160

RNU4ATAC	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU7-1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	NC	NC	Aicardi-Goutieres syndrome 9, 619487
ROBO1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ROBO2	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Vesicoureteral reflux 2, 610878
ROBO3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4	MENDELIOME	100,0%	100,0%	Aortic valve disease 3, 618496
ROGDI	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kohlschutter-Tonz syndrome, 226750
ROM1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 7, digenic form, 608133
ROR1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 108, 617654

ROR2	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	97,0%	97,0%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 42, 616622
RP1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 1, 180100
RP1L1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Occult macular dystrophy, 613587 Retinitis pigmentosa 88, 618826
RP2	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 2, 312600
RP9	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Retinitis pigmentosa 9, 180104
RPA1	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6, 619767
RPE65	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100
RPGR	VISION DISORDERS MENDELIOME	100,0%	99,7%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Macular degeneration, X-linked atrophic, 300834

RPGRIP1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	99,8%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPIA	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, 35, 300998
RPL10L	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 63, 619689
RPL11	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 7, 612562
RPL13	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RPL15	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	?Diamond-Blackfan anemia 12, 615550

RPL18	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	?Diamond-Blackfan anemia 18, 618310
RPL21	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Hypotrichosis 12, 615885
RPL26	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS	100,0%	100,0%	?Diamond-Blackfan anemia 11, 614900
RPL27	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	?Diamond-Blackfan anemia 16, 617408
RPL31	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RPL35	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	?Diamond-Blackfan anemia 19, 618312
RPL35A	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 5, 612528
RPL3L	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 2D, 619371
RPL4	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RPL5	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 6, 612561

RPL9	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RPN2	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RPS10	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 9, 613308
RPS14	MENDELIOME	100,0%	100,0%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	80,4%	80,4%	?Diamond-Blackfan anemia 20, 618313
RPS17	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 4, 612527
RPS19	HEREDITARY BONE MARROW FAILURE INTELLECTUAL DISABILITY MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 1, 105650
RPS20	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
RPS23	MENDELIOME	100,0%	100,0%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diamond-blackfan anemia 3, 610629

RPS26	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 10, 613309
RPS27	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	?Diamond-Blackfan anemia 17, 617409
RPS28	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,8%	Intellectual developmental disorder, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RPS7	HEREDITARY BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Diamond-Blackfan anemia 8, 612563
RPSA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Asplenia, isolated congenital, 271400
RRAD	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RRAGC	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
RRAS	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	No OMIM Disease ID

RRAS2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Noonan syndrome 12, 618624 Ovarian carcinoma,
RREB1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	No OMIM Disease ID
RRM1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
RRM2B	HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RRP7A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	?Microcephaly 28, primary, autosomal recessive, 619453
RS1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinoschisis, 312700
RSPH1	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	CILIOPATHIES PRIMARY IMMUNODEFICIENCIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 12, 612650

RSPO1	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anonychia congenita, 206800
RSPRY1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepiphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RTN2	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, short stature, and polymicrogyria with seizures, 614833

RUBCN	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
RUNX2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, 119600
RUSC2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 61, 617773
RXYLT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	FETAL AKINESIA (AKI) MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	99,9%	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 619542 Minicore myopathy with external ophthalmoplegia, 255320
RYR2	HEART MENDELIOME	100,0%	100,0%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 Arrhythmogenic right ventricular dysplasia 2, 600996
S1PR2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 68, 610419

SACS	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 47, 613758 Oguchi disease-1, 258100
SALL1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	100,0%	100,0%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SAMD11	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SAMD12	MENDELIOME	100,0%	100,0%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053

SAMD9L	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE NEUROPATHIES PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SAMHD1	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chylomicron retention disease, 246700
SARDH	METABOLIC DISORDERS MENDELIOME	91,4%	91,4%	No OMIM Disease ID
SARS1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SASH1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dyschromatosis universalis hereditaria 1, 127500 ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SASH3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SASS6	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 14, primary, autosomal recessive, 616402

SAT1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SATB1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Kohlschutter-Tonz syndrome-like, 619229 Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS SCHISIS	100,0%	100,0%	Glass syndrome, 612313
SBDS	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Shwachman-Diamond syndrome, 260400
SBF1	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	VISION DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lathosterolosis, 607330
SCAF4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

SCAMP5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SCAPER	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	EPILEPSY NEUROPATHIES METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Van den Ende-Gupta syndrome, 600920
SCD5	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 79, 619086
SCLT1	CILIOPATHIES MENDELIOME	95,1%	95,1%	No OMIM Disease ID
SCN10A	SKIN DISORDERS HEART NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	Episodic pain syndrome, familial, 2, 615551
SCN11A	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES HNPD MENDELIOME	100,0%	100,0%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548

SCN1A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 Migraine, familial hemiplegic, 3, 609634 Dravet syndrome, 607208 Febrile seizures, familial, 3A, 604403 Generalized epilepsy with febrile seizures plus, type 2, 604403
SCN1B	EPILEPSY HEART HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN2A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Seizures, benign familial infantile, 3, 607745 Developmental and epileptic encephalopathy 11, 613721 Episodic ataxia, type 9, 618924
SCN2B	HEART HNPD MENDELIOME	100,0%	100,0%	Atrial fibrillation, familial, 14, 615378
SCN3A	EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN3B	HEART HNPD MENDELIOME	100,0%	100,0%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4A	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Paramyotonia congenita, 168300 Hypokalemic periodic paralysis, type 2, 613345 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hyperkalemic periodic paralysis, type 2, 170500
SCN4B	HEART HNPD MENDELIOME	100,0%	100,0%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819

SCN5A	HEART MENDELIOME	100,0%	100,0%	Ventricular fibrillation, familial, 1, 603829 Heart block, progressive, type IA, 113900 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Long QT syndrome 3, 603830 Sick sinus syndrome 1, 608567 Brugada syndrome 1, 601144 Atrial fibrillation, familial, 10, 614022
SCN7A	HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SCN8A	MOVEMENT DISORDERS EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558
SCN9A	SKIN DISORDERS NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Erythermalgia, primary, 133020 Insensitivity to pain, congenital, 243000 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Neuropathy, hereditary sensory and autonomic, type IID, 243000
SCNN1A	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 2, 618114
SCO1	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 4, 619048

SCO2	VISION DISORDERS HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCUBE3	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SCYL1	NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SCYL2	FETAL AKINESIA (AKI) MENDELIOME	100,0%	100,0%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SDCCAG8	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SDHA	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165

SDHAF1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHAF2	MENDELIOME HEREDITARY CANCER	99,9%	98,4%	Paragangliomas 2, 601650
SDHB	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING HEREDITARY CANCER	80,1%	80,1%	Paragangliomas 1, with or without deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300
SDR9C7	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Craniolenticulosutural dysplasia, 607812
SEC23B	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cole-Carpenter syndrome 2, 616294
SEC31A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Halperin-Birk syndrome, 618651

SEC61A1	PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEC61B	LIVER DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SEC63	LIVER DISORDERS MENDELIOME	100,0%	100,0%	Polycystic liver disease 2, 617004
SECISBP2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Extraoral halitosis due to MTO deficiency, 618148
SELENOI	MENDELIOME	100,0%	100,0%	Spastic paraplegia 81, autosomal recessive, 618768
SELENON	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	93,0%	91,5%	Myopathy, congenital, with fiber-type disproportion, 255310 Muscular dystrophy, rigid spine, 1, 602771
SEMA3A	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SEMA3E	CRANIOFACIAL ANOMALIES HYPOGONADOTROPIC HYPOGONADISM PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	?CHARGE syndrome, 214800
SEMA4A	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283

SEMA6B	EPILEPSY MENDELIOME	100,0%	100,0%	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia type 2D, 613811
SEPTIN12	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 10, 614822
SEPTIN9	NEUROPATHIES HNPD MENDELIOME SCHISIS	100,0%	100,0%	Amyotrophy, hereditary neuralgic, 162100
SERAC1	MOVEMENT DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490
SERPINA12	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SERPINA3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Alpha-1-antichymotrypsin deficiency, Cerebrovascular disease, occlusive,
SERPINA6	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 91, 613453

SERPINB7	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peeling skin syndrome 5, 617115
SERPIN C1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thrombophilia 7 due to antithrombin III deficiency, 613118
SERPIND1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356
SERPINE1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type X, 613848
SERPINI1	EPILEPSY MENDELIOME	100,0%	100,0%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218

SET	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,4%	Intellectual developmental disorder, autosomal dominant 58, 618106
SETBP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Schinzel-Giedion midface retraction syndrome, 269150 Intellectual developmental disorder, autosomal dominant 29, 616078
SETD1A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Epilepsy, early-onset, with or without developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Luscan-Lumish syndrome, 616831
SETD5	INTELLECTUAL DISABILITY MENDELIOME	98,0%	98,0%	Intellectual developmental disorder, autosomal dominant 23, 615761
SETX	ALS MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SEZ6	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SF3B1	IRON DISORDERS MENDELIOME	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286
SF3B2	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	Craniofacial microsomia, 164210
SF3B4	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Acrofacial dysostosis 1, Nager type, 154400

SFRP4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pyle disease, 265900
SFTPA1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Interstitial lung disease 1, 619611
SFTPA2	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Interstitial lung disease 2, 178500
SFTPB	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	MENDELIOME	100,0%	100,0%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	IRON DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	MOVEMENT DISORDERS MENDELIOME	91,2%	91,2%	Dystonia-11, myoclonic, 159900
SGCG	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700

SGMS2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGO1	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT NEUROPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephrotic syndrome, type 14, 617575
SGSH	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	99,9%	99,4%	Cherubism, 118400

SH3KBP1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency 61, 300310
SH3PXD2B	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Frank-ter Haar syndrome, 249420
SH3TC2	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK2	INTELLECTUAL DISABILITY MENDELIOME	98,9%	98,9%	No OMIM Disease ID
SHANK3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	98,3%	97,3%	Phelan-McDermid syndrome, 606232
SHH	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Microphtalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHMT2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SHOC2	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome-like with loose anagen hair 1, 607721

SHOX	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	95,1%	95,1%	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300
SHROOM3	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SHROOM4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434
SI	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sucrase-isomaltase deficiency, congenital, 222900
SIAH1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Buratti-Harel syndrome, 619314
SIGLEC7	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SIGMAR1	ALS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SIK1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 30, 616341
SIK3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162

SIL1	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Marinesco-Sjogren syndrome, 248800
SIN3A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Witteveen-Kolk syndrome, 613406
SIN3B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SIPA1L3	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Cataract 45, 616851
SIX1	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT MENDELIOME SCHISIS	100,0%	100,0%	Deafness, autosomal dominant 23, 605192 Branchioototic syndrome 3, 608389
SIX3	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Schizencephaly, 269160 Holoprosencephaly 2, 157170
SIX5	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME SCHISIS	100,0%	100,0%	Branchiootorenal syndrome 2, 610896
SIX6	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Shprintzen-Goldberg syndrome, 182212

SKIV2L	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Trichohepatoenteric syndrome 2, 614602
SLC10A1	MENDELIOME	100,0%	100,0%	Hypercholanemia, familial 2, 619256
SLC10A2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Bile acid malabsorption, primary, 1, 613291
SLC10A7	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	HEARING IMPAIRMENT METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,2%	96,2%	Bartter syndrome, type 1, 601678
SLC12A2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081
SLC12A3	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Gitelman syndrome, 263800
SLC12A5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	97,4%	97,4%	Developmental and epileptic encephalopathy 34, 616645
SLC12A6	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000

SLC13A3	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	VISION DISORDERS RENAL DISORDERS MENDELIOME	100,0%	100,0%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 25, 605583
SLC17A9	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A1	MENDELIOME	100,0%	100,0%	?Megaloblastic anemia, folate-responsive, 601775

SLC19A2	HEREDITARY BONE MARROW FAILURE HEARING IMPAIRMENT IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	98,7%	98,7%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dicarboxylic aminoaciduria, 222730
SLC1A2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 41, 617105
SLC1A3	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Episodic ataxia, type 6, 612656
SLC1A4	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	MOVEMENT DISORDERS MENDELIOME PARKINSON	100,0%	100,0%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypouricemia, renal, 220150
SLC22A18	MENDELIOME	100,0%	100,0%	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210

SLC22A4	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLC22A5	HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Carnitine deficiency, systemic primary, 212140
SLC24A1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Albinism, oculocutaneous, type VI, 113750
SLC25A1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A11	MENDELIOME	100,0%	100,0%	Paragangliomas 6, 618464
SLC25A12	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471

SLC25A15	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	HEART METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	99,7%	99,7%	Fontaine progeroid syndrome, 612289
SLC25A26	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial phosphate carrier deficiency, 610773

SLC25A32	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A37	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLC25A38	HEREDITARY BONE MARROW FAILURE IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC25A42	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC26A1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Epiphyseal dysplasia, multiple, 4, 226900 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050

SLC26A3	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 61, 613865
SLC26A8	MENDELIOME	100,0%	100,0%	Spermatogenic failure 3, 606766
SLC27A4	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis prematurity syndrome, 608649
SLC28A1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLC29A3	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A10	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Arterial tortuosity syndrome, 208050

SLC2A2	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fanconi-Bickel syndrome, 227810
SLC2A9	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypouricemia, renal, 2, 612076
SLC30A10	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	MENDELIOME	100,0%	100,0%	Zinc deficiency, transient neonatal, 608118
SLC30A5	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLC30A9	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Birk-Landau-Perez syndrome, 617595
SLC33A1	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypophosphatemic rickets with hypercalciuria, 241530

SLC35A1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	81,0%	81,0%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35C1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Schneckenbecken dysplasia, 269250
SLC36A2	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240

SLC38A8	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING	93,6%	93,5%	?Hyperostosis cranialis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Acrodermatitis enteropathica, 201100
SLC39A5	VISION DISORDERS MENDELIOME	100,0%	100,0%	Myopia 24, autosomal dominant, 615946
SLC39A7	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC39A8	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,6%	96,6%	Cystinuria, 220100

SLC40A1	IRON DISORDERS LIVER DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME	100,0%	100,0%	Hemochromatosis, type 4, 606069
SLC41A1	RENAL DISORDERS MENDELIOME	100,0%	100,0%	?Nephronophthisis-like nephropathy 2, 619468
SLC44A1	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC44A4	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 72, 617606
SLC45A1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Albinism, oculocutaneous, type IV, 606574
SLC46A1	HEREDITARY BONE MARROW FAILURE IRON DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Folate malabsorption, hereditary, 229050
SLC4A1	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,1%	96,1%	Distal renal tubular acidosis 1, 179800 Spherocytosis, type 4, 612653 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900

SLC4A11	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	SKIN DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC51A	MENDELIOME	100,0%	100,0%	?Cholestasis, progressive familial intrahepatic, 6, 619484
SLC51B	MENDELIOME	100,0%	100,0%	?Bile acid malabsorption, primary, 2, 619481
SLC52A1	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Riboflavin deficiency, 615026
SLC52A2	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	MOVEMENT DISORDERS HEARING IMPAIRMENT NEUROPATHIES METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530

SLC5A1	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glucose/galactose malabsorption, 606824
SLC5A2	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Renal glucosuria, 233100
SLC5A5	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid dyshormonogenesis 1, 274400
SLC5A6	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodegeneration, infantile-onset, biotin-responsive, 618973
SLC5A7	FETAL AKINESIA (AKI) NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Myoclonic-atonic epilepsy, 616421
SLC6A17	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A2	MENDELIOME	100,0%	100,0%	?Orthostatic intolerance, 604715
SLC6A20	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500

SLC6A3	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperekplexia 3, 614618
SLC6A6	MENDELIOME	100,0%	100,0%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLC6A8	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	FETAL AKINESIA (AKI) METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 68, 615725
SLC7A6OS	MENDELIOME	100,0%	100,0%	Epilepsy, progressive myoclonic, 12, 619191
SLC7A7	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lysinuric protein intolerance, 222700
SLC7A9	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cystinuria, 220100
SLC8B1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

SLC9A1	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lichtenstein-Knorr syndrome, 616291
SLC9A3	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,4%	98,1%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	HEARING IMPAIRMENT NEUROPATHIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,7%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SLC9A7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	METABOLIC DISORDERS MENDELIOME	99,9%	99,9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLFN14	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Bleeding disorder, platelet-type, 20, 616913
SLRRP	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID

SLIT3	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLTRK1	MENDELIOME	100,0%	100,0%	Tourette syndrome, 137580 ?Trichotillomania, 613229
SLTRK6	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness and myopia, 221200
SLMAP	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SLURP1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Meleda disease, 248300
SLX4	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Fanconi anemia, complementation group P, 613951
SMAD1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SMAD2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Loeys-Dietz syndrome 6, 619656 Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Loeys-Dietz syndrome 3, 613795

SMAD4	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS HEREDITARY CANCER	100,0%	100,0%	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Aortic valve disease 2, 614823
SMAD9	HEART MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Pulmonary hypertension, primary, 2, 615342
SMARCA1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	99,9%	No OMIM Disease ID
SMARCA2	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	98,4%	98,2%	Nicolaides-Baraitser syndrome, 601358 Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Coffin-Siris syndrome 4, 614609
SMARCA5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SMARCAD1	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Basan syndrome, 129200 Huriez syndrome, 181600 Adermatoglyphia, 136000

SMARCAL1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Schimke immunoosseous dysplasia, 242900
SMARCB1	SKIN DISORDERS HNPD SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCC2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 8, 618362
SMARCD1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 11, 618779
SMARCD2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Specific granule deficiency 2, 617475
SMARCE1	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Coffin-Siris syndrome 5, 616938
SMC1A	CRANIOFACIAL ANOMALIES EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044

SMC3	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Cornelia de Lange syndrome 3, 610759
SMCHD1	MENDELIOME SCHISIS MUSCLE DISORDERS	100,0%	100,0%	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMDT1	MOVEMENT DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	No OMIM Disease ID
SMG8	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Alzahrani-Kuwahara syndrome, 619268
SMG9	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Heart and brain malformation syndrome, 616920
SMN1	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING	94,6%	94,6%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-4, 271150 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300
SMO	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia with limb anomalies, 206920
SMOC2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400

SMPD1	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SMPD4	FETAL AKINESIA (AKI) EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMPX	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Myopathy, distal, 7, adult-onset, X-linked, 301075 Deafness, X-linked 4, 300066
SMS	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAI2	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP25	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	MOVEMENT DISORDERS MENDELIOME PARKINSON	79,1%	79,1%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	MENDELIOME	100,0%	100,0%	Dementia, Lewy body, 127750

SNIP1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORA31	PRIMARY IMMUNODEFICIENCIES MENDELIOME	NC	NC	No OMIM Disease ID
SNORD11B	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 33, 610359
SNRNPB	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Cerebrocostomandibular syndrome, 117650
SNRPE	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Hypotrichosis 11, 615059
SNRPN	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Prader-Willi syndrome, 176270
SNTA1	HEART MENDELIOME	100,0%	100,0%	Long QT syndrome 12, 612955
SNX10	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 20, 616354

SNX27	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SOBP	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,7%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOCS1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SOD1	ALS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SOD2	HEART METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
SOHLH1	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	ZTTK syndrome, 617140
SORD	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	98,6%	96,1%	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912

SOS1	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Noonan syndrome 9, 616559
SOST	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	MOVEMENT DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 9, 615866

SOX17	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Vesicoureteral reflux 3, 613674
SOX18	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,3%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	VISION DISORDERS SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX3	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Coffin-Siris syndrome 10, 618506
SOX5	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Lamb-Shaffer syndrome, 616803
SOX6	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Tolchin-Le Caignec syndrome, 618971
SOX9	CRANIOFACIAL ANOMALIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290

SP110	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XII, 613849
SPAG1	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 28, 615505
SPAG17	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 55, 619380
SPAG6	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SPARC	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XVII, 616507
SPART	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Troyer syndrome, 275900
SPAST	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 6, 102530
SPATA22	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

SPATA5	HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPATA5L1	MENDELIOME	100,0%	100,0%	Deafness, autosomal recessive 119, 619615 Neurodevelopmental disorder with hearing loss and spasticity, 619616
SPATA7	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPECC1L	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	97,8%	96,2%	Teebi hypertelorism syndrome 1, 145420 ?Facial clefting, oblique, 1, 600251
SPEF2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 43, 618751
SPEG	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	99,8%	Centronuclear myopathy 5, 615959
SPEN	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Radio-Tartaglia syndrome, 619312
SPG11	ALS MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Mast syndrome, 248900

SPG7	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 7, autosomal recessive, 607259
SPI1	MENDELIOME	100,0%	100,0%	Agammaglobulinemia 10, autosomal dominant, 619707
SPIDR	MENDELIOME	100,0%	100,0%	Ovarian dysgenesis 9, 619665
SPINK1	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Tropical calcific pancreatitis, 608189 Pancreatitis, hereditary, 167800
SPINK2	MALE INFERTILITY MENDELIOME	99,3%	99,3%	?Spermatogenic failure 29, 618091
SPINK5	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Netherton syndrome, 256500
SPINT2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,6%	?Deafness, autosomal recessive 115, 618457
SPO11	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SPOCK1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SPOP	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829
SPP2	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID

SPPL2A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 86, mycobacteriosis, 619549
SPR	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY HEREDITARY CANCER	100,0%	100,0%	Legius syndrome, 611431
SPRED2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME/RASOPATHY	100,0%	100,0%	Noonan syndrome 14, 619745
SPRTN	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ruijs-Aalfs syndrome, 616200
SPRY4	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spherocytosis, type 3, 270970 Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140
SPTAN1	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 5, 613477

SPTB	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649
SPTBN1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,9%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	NEUROPATHIES HNPD METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	NEUROPATHIES HNPD METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SQOR	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	ALS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250

SRC	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
SRCAP	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140
SRD5A2	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SREBF1	MENDELIOME	96,9%	96,9%	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 Mucoepithelial dysplasia, hereditary, 158310
SRF	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SRI	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SRP54	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neutropenia, severe congenital, 8, autosomal dominant, 618752

SRP72	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Bone marrow failure syndrome 1, 614675
SRPK3	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	No OMIM Disease ID
SRPX2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SRY	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME	50,0%	50,0%	46XY sex reversal 1, 400044
SSBP1	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
SSR4	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SSX1	MENDELIOME	100,0%	100,0%	?Sarcoma, synovial, 300813
SSX2	MENDELIOME	100,0%	100,0%	?Sarcoma, synovial, 300813
ST14	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	95,8%	95,2%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090

ST3GAL5	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	98,7%	98,7%	Salt and pepper developmental regression syndrome, 609056
STAB2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
STAC3	FETAL AKINESIA (AKI) MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Myopathy, congenital, Baily-Bloch, 255995
STAG1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 47, 617635
STAG2	INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,4%	Holoprosencephaly 13, X-linked, 301043 Mullegama-Klein-Martinez syndrome, 301022
STAG3	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY	100,0%	100,0%	Spermatogenic failure 61, 619672 Premature ovarian failure 8, 615723
STAMBP	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Microcephaly-capillary malformation syndrome, 614261
STAR	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lipoid adrenal hyperplasia, 201710
STARD7	MENDELIOME	100,0%	100,0%	Epilepsy, familial adult myoclonic, 2, 607876
STAT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	95,6%	95,5%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796

STAT2	PRIMARY IMMUNODEFICIENCIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
STAT5B	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578
STAT6	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
STEAP3	IRON DISORDERS MENDELIOME	100,0%	100,0%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
CXorf56	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 7, primary, autosomal recessive, 612703

STIM1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	STING-associated vasculopathy, infantile-onset, 615934
STK11	BREAST CANCER SKIN DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Melanoma, malignant, somatic, 155600 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK36	CILIOPATHIES MENDELIOME	100,0%	100,0%	?Ciliary dyskinesia, primary, 46, 619436
STK4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	HEREDITARY BONE MARROW FAILURE LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	MENDELIOME	98,2%	96,5%	Preeclampsia/eclampsia 4, 609404

STRA6	VISION DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 16, 603720
STS	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	97,4%	97,3%	Ichthyosis, X-linked, 308100
STT3A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Pseudohypoparathyroidism, type IB, 603233
STX1B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Generalized epilepsy with febrile seizures plus, type 9, 616172

STX3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Retinal dystrophy and microvillus inclusion disease, 619446 Diarrhea 12, with microvillus atrophy, 619445
STX5	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
STXBP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Developmental and epileptic encephalopathy 4, 612164
STXBP2	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	99,8%	98,7%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	HEARING IMPAIRMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	99,9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
SUFU	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SHH MEDULLOBLASTOMA HEREDITARY CANCER	100,0%	100,0%	Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
SUGCT	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Glutaric aciduria III, 231690

SULF1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SULT2B1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	MOVEMENT DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Multiple sulfatase deficiency, 272200
SUMO1	CRANIOFACIAL ANOMALIES MENDELIOME	69,7%	69,4%	?Orofacial cleft 10, 613705
SUN5	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 16, 617187
SUOX	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sulfite oxidase deficiency, 272300
SUPT16H	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480
SURF1	HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Imagawa-Matsumoto syndrome, 618786

SVBP	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SVIL	MENDELIOME	100,0%	100,0%	Myofibrillar myopathy 10, 619040
SYCE1	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947
SYCP2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 1, 258150
SYCP3	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYK	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 82 with systemic inflammation, 619381
SYN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 50, 300115 Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
SYNE1	FETAL AKINESIA (AKI) MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	98,8%	98,8%	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	MENDELIOME	100,0%	100,0%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 76, 615540
SYNGAP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 5, 612621

SYNJ1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinson disease 20, early-onset, 615530 Developmental and epileptic encephalopathy 53, 617389
SYP	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Baker-Gordon syndrome, 618218
SYT14	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	NEUROPATHIES MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
SZT2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 18, 615476
TAB2	CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 8, 619052

TACR3	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	MOVEMENT DISORDERS CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME PARKINSON	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TAF13	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 60, 617432
TAF1A	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TAF1C	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
TAF2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 40, 615599
TAF4B	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 13, 615841
TAF6	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Alazami-Yuan syndrome, 617126
TAZ	HEREDITARY BONE MARROW FAILURE HEART PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Barth syndrome, 302060

TAL1	MENDELIOME	100,0%	100,0%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	MENDELIOME	100,0%	100,0%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Transaldolase deficiency, 606003
TANC2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration,
TAOK1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TAP1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Bare lymphocyte syndrome, type I, 604571
TAP2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571

TAPBP	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	96,6%	96,6%	Bare lymphocyte syndrome, type I, 604571
TAPT1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	ALS MENDELIOME	100,0%	100,0%	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS1	MENDELIOME	100,0%	100,0%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 21, 615918
TASP1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Suleiman-El-Hattab syndrome, 618950
TAT	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Tyrosinemia, type II, 276600
TAX1BP3	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TBC1D20	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Warburg micro syndrome 4, 615663

TBC1D23	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal dominant 65, 616044 Developmental and epileptic encephalopathy 16, 615338 DOORS syndrome, 220500
TBC1D2B	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,7%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D32	CILIOPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TBC1D7	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBC1D8B	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Nephrotic syndrome, type 20, 301028
TBCD	FETAL AKINESIA (AKI) MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	EPILEPSY NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207

TBCK	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	ALS MENDELIOME	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1X	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBL1Y	HEARING IMPAIRMENT MENDELIOME	50,0%	50,0%	?Deafness, Y-linked 2, 400047
TBP	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 17, 607136
TBR1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEART HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	98,1%	95,9%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX15	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Cousin syndrome, 260660
TBX18	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Congenital anomalies of kidney and urinary tract 2, 143400

TBX19	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Adrenocorticotropic hormone deficiency, 201400
TBX2	MENDELIOME SCHISIS	100,0%	100,0%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrial septal defect 4, 611363
TBX21	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Asthma and nasal polyps, 208550 ?Immunodeficiency 88, 619630
TBX22	CRANIOFACIAL ANOMALIES MENDELIOME SCHISIS	100,0%	100,0%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Ulnar-mammary syndrome, 181450
TBX4	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Holt-Oram syndrome, 142900
TBX6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondylocostal dysostosis 5, 122600
TBXA2R	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	99,7%	No OMIM Disease ID

TBXAS1	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ghosal hematodiaphyseal syndrome, 231095
TBXT	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sacral agenesis with vertebral anomalies, 615709
TCAP	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	CRANIOFACIAL ANOMALIES DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Craniosynostosis 3, 615314 Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCF20	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Agammaglobulinemia 8B, autosomal recessive, 619824 Agammaglobulinemia 8A, autosomal dominant, 616941
TCF4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TCHH	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Uncombable hair syndrome 3, 617252

TCIRG1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Transcobalamin II deficiency, 275350
TCOF1	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Treacher Collins syndrome 1, 154500
TCTN1	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	95,5%	94,7%	Joubert syndrome 13, 614173
TCTN2	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885

TCTN3	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDGF1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Forebrain defects,
TDP1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cataract 36, 613887
TDRD9	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 30, 618110
TDRKH	NEUROPATHIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TEAD1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Sveinsson chorioretinal atrophy, 108985

TECPR2	MOVEMENT DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 14, 614020
TECRL	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	VISION DISORDERS SKIN DISORDERS MENDELIOME	100,0%	100,0%	Venous malformations, multiple cutaneous and mucosal, 600195 Glaucoma 3, primary congenital, E, 617272
TELO2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	You-Hoover-Fong syndrome, 616954
TENM3	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TENM4	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Essential tremor, hereditary, 5, 616736
TENT5A	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XVIII, 617952
TERB1	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 60, 619646

TERB2	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 59, 619645
TERC	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS MENDELIOME HEREDITARY CANCER	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME HEREDITARY CANCER	83,7%	83,7%	No OMIM Disease ID
TERT	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS MELANOMA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TES	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TET2	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
TET3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Beck-Fahrner syndrome, 618798
TEX11	MALE INFERTILITY MENDELIOME	97,1%	97,1%	Spermatogenic failure, X-linked 2, 309120

TEX14	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 23, 617707
TEX15	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 25, 617960
TF	IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Atransferrinemia, 209300
TFAM	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Branchiooculofacial syndrome, 113620
TFAP2B	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Patent ductus arteriosus 2, 617035 Char syndrome, 169100
TFB2M	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
TFE3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, with pigmentary mosaicism and coarse facies, 301066 Renal cell carcinoma, papillary, 1, 300854
TFG	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spastic paraparesis 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFPT	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TFR2	IRON DISORDERS LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemochromatosis, type 3, 604250

TFRC	IRON DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 46, 616740
TG	MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Thyroid dyshormonogenesis 3, 274700
TGDS	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Catel-Manzke syndrome, 616145
TGFB1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
TGFB2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Loeys-Dietz syndrome 4, 614816
TGFB3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	VISION DISORDERS MENDELIOME	100,0%	100,0%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471

TGFB1	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	99,9%	Loeys-Dietz syndrome 1, 609192
TGFB2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
TGIF1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Holoprosencephaly 4, 142946
TGM1	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Uncombable hair syndrome 2, 617251
TGM5	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Peeling skin syndrome 2, 609796
TGM6	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 35, 613908
TH	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY NEWBORN BLOOD SPOT SCREENING MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Segawa syndrome, recessive, 605407

THAP1	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Dystonia 6, torsion, 602629
THBD	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	100,0%	100,0%	Thrombophilia 12 due to thrombomodulin defect, 614486
THBS4	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
THG1L	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC1	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
THOC2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 12, 300957
THOC6	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Thrombocythemia 1, 187950
THRA	MENDELIOME	100,0%	100,0%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRΒ	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650
THSD1	MENDELIOME	100,0%	100,0%	?Aneurysm, intracranial berry, 12, 618734

THSD4	MENDELIOME	100,0%	100,0%	Aortic aneurysm, familial thoracic 12, 619825
THUMPD1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TIA1	MENDELIOME	100,0%	100,0%	Welander distal myopathy, 604454 Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133
TICAM1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TIE1	MENDELIOME	100,0%	100,0%	Lymphatic malformation 11, 619401
TIMM22	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
TIMM50	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Mohr-Tranebjærg syndrome, 304700
TIMMDC1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	VISION DISORDERS MENDELIOME	100,0%	100,0%	Sorsby fundus dystrophy, 136900

TINF2	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TJP1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TJP2	HEARING IMPAIRMENT LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING	98,8%	98,8%	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TK2	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	98,8%	98,7%	Short stature, developmental delay, and congenital heart defects, 617044
TLCD3B	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cone-rod dystrophy 22, 619531
TLE6	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Preimplantation embryonic lethality, 616814

TLK2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 57, 618050
TLL1	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Atrial septal defect 6, 613087
TLR3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TLR4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TLR5	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TLR7	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TMC1	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermodysplasia verruciformis, 226400
TMC8	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Epidermodysplasia verruciformis 2, 618231

TMCO1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	88,0%	88,0%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMCO3	VISION DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TMEM106B	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM126A	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Optic atrophy 7, 612989
TMEM126B	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM127	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
TMEM132E	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 99, 618481
TMEM138	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 16, 614465
TMEM14C	IRON DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID

TMEM165	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIk, 614727
TMEM186	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
TMEM199	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM218	VISION DISORDERS CILIOPATHIES MENDELIOME	100,0%	100,0%	Joubert syndrome 39, 619562
TMEM222	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397

TMEM237	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Joubert syndrome 14, 614424
TMEM240	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 21, 607454
TMEM251	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Dysostosis multiplex, Ain-Naz type, 619345
TMEM260	CONGENITAL HEART DISEASE CILIOPATHIES HEART RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	HEARING IMPAIRMENT HEART MENDELIOME	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 5, 604400 Auditory neuropathy, autosomal dominant 3, 619832 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM53	MENDELIOME	100,0%	100,0%	Craniotubular dysplasia, Ikegawa type, 619727
TMEM63A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM65	MENDELIOME MITOCHONDRIAL DISORDERS	99,8%	97,9%	No OMIM Disease ID

TMEM67	MOVEMENT DISORDERS VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TMEM70	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	VISION DISORDERS MENDELIOME	100,0%	100,0%	Nanophthalmos 4, 615972
TMIE	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 6, 600971
TMLHE	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	99,6%	99,5%	No OMIM Disease ID
TMPO	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TMPRSS15	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Enterokinase deficiency, 226200
TMPRSS3	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Iron-refractory iron deficiency anemia, 206200

TMTC2	HEARING IMPAIRMENT MENDELIOME	97,5%	97,5%	No OMIM Disease ID
TMTC3	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Lissencephaly 8, 617255
TMX2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNC	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Deafness, autosomal dominant 56, 615629
TNFAIP3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TNFRSF10B	MENDELIOME	100,0%	100,0%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	99,7%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF11B	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	92,8%	92,8%	Periodic fever, familial, 142680

TNFRSF4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Immunodeficiency 16, 615593
TNFRSF9	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TNFSF11	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TNFSF13	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TNIK	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 54, 617028
TNNC1	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	FETAL AKINESIA (AKI) MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Arthrogryposis, distal, type 2B1, 601680
TNNI3	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	CONGENITAL HEART DISEASE HEART MENDELIOME	100,0%	100,0%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Nemaline myopathy 5, Amish type, 605355

TNNT2	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TNNT3	FETAL AKINESIA (AKI) MENDELIOME	100,0%	100,0%	Arthrogryposis, distal, type 2B2, 618435
TNPO2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNPO3	MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNR	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNRC6A	MENDELIOME	100,0%	100,0%	?Epilepsy, familial adult myoclonic, 6, 618074
TNRC6B	INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Global developmental delay with speech and behavioral abnormalities, 619243
TNS1	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TNS2	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TNXB	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 7, 614969

TOGARAM1	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Joubert syndrome 37, 619185
TOM1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Immunodeficiency 85 and autoimmunity, 619510
TOMM70	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
TONSL	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spondyloepiphyseal dysplasia, sponastrime type, 271510
TOP1	MENDELIOME	100,0%	100,0%	DNA topoisomerase I, camptothecin-resistant,
TOP2A	MENDELIOME	100,0%	100,0%	DNA topoisomerase II, resistance to inhibition of, by amsacrine,
TOP2B	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TOP3A	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TOPORS	VISION DISORDERS CILIOPATHIES MENDELIOME	100,0%	100,0%	Retinitis pigmentosa 31, 609923
TOR1A	FETAL AKINESIA (AKI) MOVEMENT DISORDERS HNPD INTELLECTUAL DISABILITY MENDELIOME	92,9%	91,5%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TOR1AIP1	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072

TP53	HEREDITARY BONE MARROW FAILURE BREAST CANCER MENDELIOME SHH MEDULLOBLASTOMA HEREDITARY CANCER	91,7%	91,7%	Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Nasopharyngeal carcinoma, somatic, 607107 Bone marrow failure syndrome 5, 618165
TP53RK	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galloway-Mowat syndrome 4, 617730
TP63	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Split-hand/foot malformation 4, 605289 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285 Limb-mammary syndrome, 603543
TP73	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPCN2	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TPI1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	METABOLIC DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	HEART MENDELIOME	100,0%	100,0%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878

TPM2	FETAL AKINESIA (AKI) MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPM4	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TPMT	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TPO	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Thyroid dyshormonogenesis 2A, 274500
TPP1	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TPRKB	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	82,3%	81,9%	Galloway-Mowat syndrome 5, 617731
TPRN	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	97,6%	96,0%	Deafness, autosomal recessive 79, 613307
TRAC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387

TRAF3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TRAF3IP1	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Senior-Loken syndrome 9, 616629
TRAF3IP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	?Candidiasis, familial, 8, 615527
TRAF6	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TRAF7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Seckel syndrome 9, 616777
TRAK1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
MAP11	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Microcephaly 25, primary, autosomal recessive, 618351

TRAPPC2	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 13, 613192
TRDN	HEART MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TREH	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	Trehalase deficiency, 612119
TREM2	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193

TREX1	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRH	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
TRHR	MENDELIOME	100,0%	100,0%	Hypothyroidism, congenital, nongoitrous, 7, 618573
TRIM2	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	93,9%	93,9%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM22	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TRIM28	MENDELIOME HEREDITARY CANCER	100,0%	100,0%	No OMIM Disease ID
TRIM32	VISION DISORDERS CILIOPATHIES SKIN DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Anencephaly 1, 206500
TRIM37	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS HEREDITARY CANCER	98,7%	98,7%	Mulibrey nanism, 253250

TRIM44	MENDELIOME	100,0%	100,0%	?Aniridia 3, 617142
TRIM63	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TRIM71	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Hydrocephalus, congenital communicating, 1, 618667
TRIM8	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	INTELLECTUAL DISABILITY MENDELIOME	99,9%	99,7%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIOBP	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 28, 609823
TRIP11	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Odontochondrodysplasia 1, 184260 Achondrogenesis, type IA, 200600
TRIP12	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIP13	MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Oocyte maturation defect 9, 619011 Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	FETAL AKINESIA (AKI) MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 68, 618302

TRMT10A	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	LIVER DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Liver failure, transient infantile, 613070
TRNT1	VISION DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPA1	HNPD MENDELIOME	100,0%	100,0%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	MENDELIOME	100,0%	100,0%	?Spinocerebellar ataxia 41, 616410
TRPC6	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM3	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID

TRPM4	SKIN DISORDERS HEART MENDELIOME	100,0%	100,0%	Progressive familial heart block, type IB, 604559 Erythrokeratoderma variabilis et progressiva 6, 618531
TRPM6	EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypomagnesemia 1, intestinal, 602014
TRPM8	HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TRPS1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Trichorhinophalangeal syndrome, type III, 190351 Trichorhinophalangeal syndrome, type I, 190350
TRPV1	HNPD MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TRPV3	SKIN DISORDERS HNPD MENDELIOME	97,1%	97,1%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome 1, 614594
TRPV4	FETAL AKINESIA (AKI) NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 SED, Maroteaux type, 184095 Metatropic dysplasia, 156530 Scapuloperoneal spinal muscular atrophy, 181405 Hereditary motor and sensory neuropathy, type IIc, 606071 ?Avascular necrosis of femoral head, primary, 2, 617383 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500
TRPV6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	Hyperparathyroidism, transient neonatal, 618188

TRRAP	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Deafness, autosomal dominant 75, 618778 Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TSEN15	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	94,9%	94,9%	Combined oxidative phosphorylation deficiency 3, 610505

TSGA10	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 26, 617961
TSHB	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, Thyroid carcinoma with thyrotoxicosis, somatic,
TSHZ1	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Aural atresia, congenital, 607842
TSPAN12	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Exudative vitreoretinopathy 5, 613310
TSPAN7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 58, 300210
TSPEAR	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	MOVEMENT DISORDERS CILIOPATHIES MENDELIOME	100,0%	100,0%	Spinocerebellar ataxia 11, 604432
TTC12	MENDELIOME	100,0%	100,0%	Ciliary dyskinesia, primary, 45, 618801

TTC19	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	MENDELIOME	100,0%	100,0%	Spermatogenic failure 37, 618429
TTC21B	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC26	CILIOPATHIES MENDELIOME	100,0%	100,0%	Biliary, renal, neurologic, and skeletal syndrome, 619534
TTC29	MALE INFERTILITY MENDELIOME	100,0%	100,0%	Spermatogenic failure 42, 618745
TTC37	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Trichohepatoenteric syndrome 1, 222470
TTC5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC7A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Gastrointestinal defects and immunodeficiency syndrome, 243150

TTC8	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 39, 615541
TTLL5	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cone-rod dystrophy 19, 615860
TTN	FETAL AKINESIA (AKI) HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, familial hypertrophic, 9, 613765 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, dilated, 1G, 604145 Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TTPA	MOVEMENT DISORDERS NEWBORN BLOOD SPOT SCREENING MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ataxia with isolated vitamin E deficiency, 277460
TTR	HEART NEUROPATHIES HNPD MENDELIOME	94,6%	94,6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUB	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Retinal dystrophy and obesity, 616188

TUBA1A	FETAL AKINESIA (AKI) MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Lissencephaly 3, 611603
TUBA3D	VISION DISORDERS MENDELIOME	100,0%	99,9%	Keratoconus 9, 617928
TUBA4A	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	No OMIM Disease ID
TUBB	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	99,8%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	100,0%	100,0%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	FETAL AKINESIA (AKI) EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039

TUBB4A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99,5%	97,4%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	MENDELIOME	100,0%	100,0%	?Facial palsy, congenital, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	MENDELIOME	100,0%	100,0%	Oocyte maturation defect 2, 616780
TUBG1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	97,0%	97,0%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132

TUSC3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST1	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWIST2	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY MUSCLE DISORDERS	100,0%	100,0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXN2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Burn-McKeown syndrome, 608572
TXNRD2	DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME	100,0%	100,0%	?Glucocorticoid deficiency 5, 617825

TYK2	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency 35, 611521
TYMP	NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	METABOLIC DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
TYR	VISION DISORDERS SKIN DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100
TYROBP	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Albinism, oculocutaneous, type III, 203290
U2AF2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
UBA1	FETAL AKINESIA (AKI) HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME MUSCLE DISORDERS	100,0%	99,8%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054

UBA5	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBAP1	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Spastic paraplegia 80, autosomal dominant, 618418
UBB	CRANIOFACIAL ANOMALIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
UBE2A	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Nascimento type, 300860
UBE2T	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fanconi anemia, complementation group T, 616435
UBE3A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Angelman syndrome, 105830
UBE3B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Kaufman oculocerebrofacial syndrome, 244450
UBE4A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBIAD1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	ALS MENDELIOME	100,0%	100,0%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857

UBR1	SKIN DISORDERS LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	98,0%	98,0%	Johanson-Blizzard syndrome, 243800
UBR2	MALE INFERTILITY MENDELIOME	100,0%	99,9%	No OMIM Disease ID
UBR7	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Li-Campeau syndrome, 619189
UBTF	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 79, autosomal recessive, 615491
UFC1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
UGDH	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Developmental and epileptic encephalopathy 84, 618792

UGP2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	96,6%	96,3%	Developmental and epileptic encephalopathy 83, 618744
UGT1A1	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
UMOD	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UMPS	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	97,0%	97,0%	Orotic aciduria, 258900
UNC119	MENDELIOME	100,0%	100,0%	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy,
UNC13A	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
UNC13D	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	MENDELIOME	100,0%	100,0%	Osteootohepatoenteric syndrome, 619377
UNC45B	VISION DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	?Cataract 43, 616279 Myofibrillar myopathy 11, 619178
UNC80	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNC93B1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	No OMIM Disease ID
UNG	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency with hyper IgM, type 5, 608106

UPB1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Beta-ureidopropionase deficiency, 613161
UPF1	INTELLECTUAL DISABILITY MENDELIOME	99,6%	99,0%	No OMIM Disease ID
UPF3B	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UPK3A	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
UQCC1	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
UQCC2	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
UQCR11	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
UQCRB	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	Parkinsonism with polyneuropathy, 619279
UQCRC2	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRCFS1	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 10, 618775

UQCRH	MENDELIOME MITOCHONDRIAL DISORDERS	100,0%	100,0%	No OMIM Disease ID
UQCRQ	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Urocanase deficiency, 276880
UROD	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Porphyria, congenital erythropoietic, 263700
USB1	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Poikiloderma with neutropenia, 604173
USH1C	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Usher syndrome, type 1G, 606943
USH2A	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	99,5%	99,5%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809

USP18	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pseudo-TORCH syndrome 2, 617397
USP26	MALE INFERTILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
USP27X	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 105, 300984
USP45	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Leber congenital amaurosis 19, 618513
USP48	HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	No OMIM Disease ID
USP53	MENDELIOME	100,0%	100,0%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658
USP7	INTELLECTUAL DISABILITY MENDELIOME	94,8%	94,8%	Hao-Fountain syndrome, 616863
USP8	MENDELIOME	100,0%	100,0%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
UST	MENDELIOME	100,0%	100,0%	No OMIM Disease ID
UVSSA	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	UV-sensitive syndrome 3, 614640
VAC14	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600

VAMP2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VANGL1	MENDELIOME	100,0%	100,0%	Caudal regression syndrome, 600145
VANGL2	MENDELIOME	100,0%	100,0%	Neural tube defects, 182940
VAPB	ALS MENDELIOME	100,0%	100,0%	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VARS1	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Combined oxidative phosphorylation deficiency 20, 615917
VAV1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	97,1%	97,1%	No OMIM Disease ID
VAX1	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING	99,9%	99,5%	?Microphthalmia, syndromic 11, 614402
VCAN	VISION DISORDERS MENDELIOME	100,0%	100,0%	Wagner syndrome 1, 143200
VCL	HEART MENDELIOME	100,0%	100,0%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	ALS MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320

VDR	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	99,9%	98,7%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	SKIN DISORDERS MENDELIOME	100,0%	100,0%	Lymphatic malformation 4, 615907
VHL	CILIOPATHIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
VIM	VISION DISORDERS MENDELIOME	100,0%	100,0%	Cataract 30, pulverulent, 116300
VIPAS39	FETAL AKINESIA (AKI) HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	93,1%	93,0%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Myopathy, X-linked, with excessive autophagy, 310440

VPS11	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Choreoacanthocytosis, 200150
VPS13B	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	99,5%	99,4%	Cohen syndrome, 216550
VPS13C	MENDELIOME PARKINSON PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Dystonia 30, 619291
VPS33A	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	89,9%	89,9%	Mucopolysaccharidosis-plus syndrome, 617303

VPS33B	FETAL AKINESIA (AKI) SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	MENDELIOME PARKINSON	100,0%	100,0%	No OMIM Disease ID
VPS35L	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraparesis 53, autosomal recessive, 614898
VPS41	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Spinocerebellar atrophy, autosomal recessive 29, 619389
VPS45	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	95,3%	95,3%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	HEREDITARY BONE MARROW FAILURE INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	CIMDAG syndrome, 619273
VPS50	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS51	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Pontocerebellar hypoplasia, type 13, 618606

VPS53	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	99,8%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	VISION DISORDERS MENDELIOME	100,0%	100,0%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microphthalmia, isolated 2, 610093 Microphthalmia with coloboma 3, 610092
VWA1	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Neuropathy, hereditary motor, with myopathic features, 619216
VWA3B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WAC	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Desanto-Shinawi syndrome, 616708
WARS1	NEUROPATHIES MENDELIOME	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	Parkinsonism-dystonia 3, childhood-onset, 619738 Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710

WAS	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	100,0%	100,0%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WASF1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 43, 615817
WASHC5	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP11	MENDELIOME	100,0%	100,0%	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227
WBP2	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 107, 617639
WDFY3	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	98,1%	98,1%	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550

WDR11	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
WDR19	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR26	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	97,0%	94,3%	Skraban-Deardorff syndrome, 617616
WDR35	CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR36	VISION DISORDERS MENDELIOME	100,0%	100,0%	Glaucoma 1, open angle, G, 609887
WDR37	INTELLECTUAL DISABILITY MENDELIOME	86,5%	86,5%	Neurooculocardiogenitourinary syndrome, 618652

WDR4	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON	100,0%	100,0%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	FETAL AKINESIA (AKI) INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,9%	96,9%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	MOVEMENT DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Galloway-Mowat syndrome 1, 251300
WDR81	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Oocyte maturation defect 5, 617996
WFS1	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300

WHRN	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Wiskott-Aldrich syndrome 2, 614493
WIPI2	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WLS	MENDELIOME	100,0%	100,0%	Zaki syndrome, 619648
WNK1	NEUROPATHIES HNPD RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	RENAL DISORDERS MENDELIOME	100,0%	100,0%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Osteogenesis imperfecta, type XV, 615220
WNT10A	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300
WNT2B	MENDELIOME	100,0%	100,0%	Diarrhea 9, 618168

WNT3	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Tetra-amelia syndrome 1, 273395
WNT4	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	99,8%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS	100,0%	100,0%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME	100,0%	100,0%	No OMIM Disease ID
WNT7A	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DKC PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Werner syndrome, 277700

WT1	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME HEREDITARY CANCER	97,7%	97,7%	Mesothelioma, somatic, 156240 Meacham syndrome, 608978 Frasier syndrome, 136680 Nephrotic syndrome, type 4, 256370 Denys-Drash syndrome, 194080 Wilms tumor, type 1, 194070
WWOX	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Xanthinuria, type I, 278300
XIAP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIRP2	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
XIST	MENDELIOME	NC	NC	X-inactivation, familial skewed, 300087
XK	MOVEMENT DISORDERS EPILEPSY HEART MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, group A, 278700
XPC	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING HEREDITARY CANCER	100,0%	100,0%	Xeroderma pigmentosum, group C, 278720

XPNPEP3	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis-like nephropathy 1, 613159
XPO5	RENAL DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
XPR1	MOVEMENT DISORDERS MENDELIOME PARKINSON	100,0%	100,0%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	HEREDITARY BONE MARROW FAILURE BREAST CANCER MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
XRCC4	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	99,7%	Desbuquois dysplasia 2, 615777

XYLT2	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING	96,7%	96,7%	Spondyloocular syndrome, 605822
YAP1	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME	100,0%	100,0%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS1	VISION DISORDERS NEUROPATHIES MENDELIOME	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS	100,0%	100,0%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YEATS2	MENDELIOME	100,0%	100,0%	?Epilepsy, myoclonic, familial adult, 4, 615127
YIF1B	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	90,1%	90,1%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING	100,0%	100,0%	?Optic atrophy 11, 617302
YPEL2	VISION DISORDERS MENDELIOME	100,0%	99,9%	No OMIM Disease ID

YRDC	MENDELIOME	100,0%	100,0%	Galloway-Mowat syndrome 10, 619609
YWHAE	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
YWHAG	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Developmental and epileptic encephalopathy 56, 617665
YWHAZ	SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
YY1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Gabriele-de Vries syndrome, 617557
YY1AP1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Grange syndrome, 602531
ZAP70	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)	100,0%	100,0%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Leukemia, acute promyelocytic, PL2F/RARA type,
ZBTB17	HEART MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ZBTB18	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 22, 612337

ZBTB20	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Primrose syndrome, 259050
ZBTB24	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Lethal congenital contracture syndrome 6, 616248
ZBTB7A	MENDELIOME	100,0%	100,0%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769
ZC3H14	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	FETAL AKINESIA (AKI) MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	100,0%	100,0%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZCCHC8	HEREDITARY BONE MARROW FAILURE MENDELIOME	100,0%	100,0%	?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674
ZDHHC9	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Raymond type, 300799
ZEB1	VISION DISORDERS MENDELIOME	100,0%	100,0%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	CRANIOFACIAL ANOMALIES EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS	97,4%	97,4%	Mowat-Wilson syndrome, 235730

ZFHX2	HNPD MENDELIOME	100,0%	100,0%	?Marsili syndrome, 147430
ZFHX3	MENDELIOME	100,0%	100,0%	Prostate cancer, somatic, 176807
ZFHX4	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ZFP57	MENDELIOME	100,0%	100,0%	Diabetes mellitus, transient neonatal 1, 601410
ZFPM2	CONGENITAL HEART DISEASE DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME	100,0%	100,0%	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500
ZFYVE26	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	MOVEMENT DISORDERS MENDELIOME	100,0%	100,0%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	?Craniosynostosis 6, 616602 Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS	100,0%	100,0%	Holoprosencephaly 5, 609637
ZIC3	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME SCHISIS	100,0%	100,0%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMIZ1	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659

ZMPSTE24	FETAL AKINESIA (AKI) SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS	100,0%	100,0%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210
ZMYM2	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522
ZMYND10	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZMYND15	MALE INFERTILITY MENDELIOME	100,0%	100,0%	?Spermatogenic failure 14, 615842
ZNF141	MENDELIOME	100,0%	100,0%	?Polydactyly, postaxial, type A6, 615226
ZNF142	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	INTELLECTUAL DISABILITY MENDELIOME	99,6%	99,6%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282

ZNF407	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	SIMHA syndrome, 619557
ZNF408	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF41	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ZNF423	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF462	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Weiss-Kruszka syndrome, 618619
ZNF469	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Brittle cornea syndrome 1, 229200
ZNF513	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	?Retinitis pigmentosa 58, 613617
ZNF526	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ZNF592	MOVEMENT DISORDERS SKIN DISORDERS MENDELIOME	100,0%	100,0%	No OMIM Disease ID
ZNF644	VISION DISORDERS MENDELIOME	100,0%	100,0%	Myopia 21, autosomal dominant, 614167
ZNF687	MENDELIOME	100,0%	100,0%	Paget disease of bone 6, 616833
ZNF699	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	DEGCAGS syndrome, 619488

ZNF711	INTELLECTUAL DISABILITY MENDELIOME	100,0%	100,0%	Intellectual developmental disorder, X-linked 97, 300803
ZNF750	SKIN DISORDERS MENDELIOME	100,0%	100,0%	?Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNFX1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	100,0%	100,0%	Immunodeficiency 91 and hyperinflammation, 619644
ZNHIT3	MENDELIOME PRE CONCEPTION SCREENING	75,8%	74,4%	PEHO syndrome, 260565
ZP1	MENDELIOME PRE CONCEPTION SCREENING	100,0%	100,0%	Oocyte maturation defect 1, 615774
ZP2	MENDELIOME	100,0%	100,0%	Oocyte maturation defect 6, 618353
ZP3	MENDELIOME	100,0%	100,0%	Oocyte maturation defect 3, 617712
ZPB1	MENDELIOME	100,0%	100,0%	?Spermatogenic failure 66, 619799
ZPR1	MENDELIOME	100,0%	100,0%	?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321
ZSWIM6	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS	97,6%	96,3%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 Acromelic frontonasal dysostosis, 603671

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

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TWIST is the chemistry used for WES analysis.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with coverage denoting NC are non-protein coding genes.

non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 19th, 2022.

This list is accurate for panel version DG 3.4.0

Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors