PRECONCEPTION SCREENING GENE PANEL DG 2.15 (1924 genes)

Releasedate: 31-01-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	106.4	100	99.7	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS	124.3	100	99.6	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	126.2	100	99.3	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	92.7	100	99.5	GABA-transaminase deficiency, 613163
ABCA1	123.6	100	99.5	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	140	99.6	97.8	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCA3	124	99.9	99.3	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	127.1	100	99.5	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2}, 153800
ABCB11	158.2	99.9	99.1	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	129.8	99.9	97.9	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCC2	135.8	100	100	Dubin-Johnson syndrome, 237500
ABCC6	116.4	93.6	92.6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	146.6	100	99.9	Diabetes mellitus, noninsulin-dependent, 125853

				Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD3	93.7	95.2	89.5	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	143.6	99.9	98.3	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	145.2	100	99.2	Sitosterolemia, 210250
ABCG8	148.4	99.2	96.6	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	107	97.3	88	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	209.6	100	99.9	Chanarin-Dorfman syndrome, 275630
ACAD8	141.5	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	101.3	98.8	95.6	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	123.9	99.3	97.6	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	119.1	99.5	95.6	2-methylbutyrylglycinuria, 610006
ACADVL	118.8	98.7	95.1	VLCAD deficiency, 201475
ACAN	121.6	91.6	85	?Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800
ACAT1	123.7	99.2	94.6	Alpha-methylacetoacetic aciduria, 203750
ACE	120.7	99.5	97.4	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase], 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 {SARS, progression of}, 0 {Stroke, hemorrhagic}, 614519
ACER3	105.8	99.9	97.5	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	129.3	95.8	91.8	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	155.3	100	100	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	196.2	100	99.9	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265

ACTA1	99.7	99.2	95.3	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACY1	132.8	99.9	98.3	Aminoacylase 1 deficiency, 609924
ADA	113	98.9	97.3	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	101.4	99.9	99.1	?Sneddon syndrome, 182410 Polyarteritis nodosa, childhood-onset, 615688
ADAM22	140.5	99.9	98.6	?Epileptic encephalopathy, early infantile, 61, 617933
ADAM9	146.3	98.6	94.1	Cone-rod dystrophy 9, 612775
ADAMTS10	107.8	99.9	98.7	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	99.9	96.3	91.7	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	117.1	88.9	86.7	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	147.5	99.9	98.9	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	117.4	98.5	96.6	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTSL2	112.2	96.5	91	Geleophysic dysplasia 1, 231050
ADAMTSL4	90.6	99.9	98.8	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	125	100	99.8	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	82.1	99.5	97.3	Mental retardation, autosomal recessive 36, 615286
ADD3	169.7	100	99.7	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	149.7	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG6	147.6	99.8	98	Lethal congenital contracture syndrome 9, 616503
ADGRV1	140.3	99.5	97	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADK	100.4	99.5	96.1	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	183.6	99.2	99.1	Adenylosuccinase deficiency, 103050
ADSSL1	113.6	89.8	85.9	Myopathy, distal, 5, 617030
AFG3L2	121	91.9	84.9	Spastic ataxia 5, autosomal recessive, 614487

				Spinocerebellar ataxia 28, 610246
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGBL5	111	100	99.8	Retinitis pigmentosa 75, 617023
AGK	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	51.7	96.8	84.8	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	114.8	95.2	89.3	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	214.2	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	134.6	92	91.9	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	139.5	99.9	99.2	Hyperoxaluria, primary, type 1, 259900
AHCY	124.5	100	99.8	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	139.3	99.2	95.1	Joubert syndrome 3, 608629
AICDA	139	89.8	82.6	Immunodeficiency with hyper-IgM, type 2, 605258
AIMP1	84.8	97.3	89.7	Leukodystrophy, hypomyelinating, 3, 260600
AIPL1	116	100	99.5	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	68.2	98.9	92	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	119.8	100	99.2	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	111.8	99.8	96.6	Reticular dysgenesis, 267500
AKR1C2	179.5	96.3	89.6	46XY sex reversal 8, 614279
AKR1D1	106.1	98.5	94.3	Bile acid synthesis defect, congenital, 2, 235555
ALAD	100.6	99.8	97.4	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALB	156	99.9	98.7	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	131.1	100	99.9	Cutis laxa, autosomal dominant 3, 616603

				Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH1A3	104.7	93.4	89.6	Microphthalmia, isolated 8, 615113
ALDH3A2	125.7	95.3	94.6	Sjogren-Larsson syndrome, 270200
ALDH4A1	116	100	98.6	Hyperprolinemia, type II, 239510
ALDH5A1	87.6	86.4	80.1	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	127.3	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	77.1	93.7	85.4	Epilepsy, pyridoxine-dependent, 266100
ALDOA	139.1	76.3	74.7	Glycogen storage disease XII, 611881
ALDOB	165.7	100	99.4	Fructose intolerance, hereditary, 229600
ALG1	50.9	53.6	48.8	Congenital disorder of glycosylation, type Ik, 608540
ALG11	139.6	96.7	96	Congenital disorder of glycosylation, type Ip, 613661
ALG12	156.2	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG2	115.9	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.4	96	93.3	Congenital disorder of glycosylation, type Ic, 603147
ALG8	126	96.5	95.1	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	124.3	100	99.6	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ALOX12B	130.6	100	99.5	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	122.2	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	156.4	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	170.2	99.9	99.2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	153.2	99.9	98.4	?Frontonasal dysplasia 3, 613456
ALX3	102.7	73.3	70.9	Frontonasal dysplasia 1, 136760

ALX4	132.7	98.4	92.5	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	157.9	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	175.1	94.6	88.5	Amelogenesis imperfecta, type IF, 616270
AMN	66.8	83.5	71.6	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	126.7	100	99.9	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	135.5	99.9	99.2	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	173.1	100	100	Glycine encephalopathy, 605899
ANGPTL3	89.9	97.5	92.2	Hypobetalipoproteinemia, familial, 2, 605019
ANK3	155.1	99.1	98.8	?Mental retardation, autosomal recessive, 37, 615493
ANKH	118.6	100	99.7	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	162.5	98	94.7	?Microcephaly 16, primary, autosomal recessive, 616681
ANKS6	91.8	92.8	88.6	Nephronophthisis 16, 615382
ANO10	116.7	98.8	96.5	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO5	142.2	99.5	95.9	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANO6	137.4	98	92.9	Scott syndrome, 262890
ANTXR1	123	98.3	95.7	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	100	98.9	94.9	Hyaline fibromatosis syndrome, 228600
AP1S1	111.3	99.9	99.5	MEDNIK syndrome, 609313
AP3B1	95	97.8	90.2	Hermansky-Pudlak syndrome 2, 608233
AP3B2	135.1	97.5	94.2	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	121	98.1	97.8	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	147.4	100	99.8	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	98.7	99.7	97.9	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	127.2	99.1	96.4	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	65.8	71.8	69.3	Spastic paraplegia 52, autosomal recessive, 614067

AP5Z1	96.8	99.7	96.6	Spastic paraplegia 48, autosomal recessive, 613647
APC2	63.5	93.3	85.3	?Sotos syndrome 3, 617169
APOB	181.7	99.6	99.3	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	99.4	100	100	Hyperlipoproteinemia, type Ib, 207750
APOE	56.2	93.9	83.1	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOPT1	63.8	81.4	78.1	Mitochondrial complex IV deficiency, 220110
APRT	68.2	100	98.7	Adenine phosphoribosyltransferase deficiency, 614723
APTX	118.9	94.2	91.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	117.4	99.5	95.6	Diabetes insipidus, nephrogenic, 125800
ARFGEF2	154.9	100	99.3	Periventricular heterotopia with microcephaly, 608097
ARG1	167.8	100	100	Argininemia, 207800
ARHGDIA	143.9	100	99.9	Nephrotic syndrome, type 8, 615244
ARL13B	97.3	98.9	92.8	Joubert syndrome 8, 612291
ARL2BP	66.3	88.3	79.3	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	85.2	99.8	95.3	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	129.1	94.4	93.4	Ciliary dyskinesia, primary, 23, 615451
ARSA	97.8	100	99.7	Metachromatic leukodystrophy, 250100
ARSB	117.5	94.9	87.7	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARV1	133.5	100	99.3	Epileptic encephalopathy, early infantile, 38, 617020
ASAH1	105.9	97.6	92.1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	114.4	99.9	98.8	Argininosuccinic aciduria, 207900
ASNS	105.8	97.8	90.5	Asparagine synthetase deficiency, 615574
ASPA	127.6	99.1	95.8	Canavan disease, 271900
ASPH	117.3	98.8	93.9	Traboulsi syndrome, 601552
ASPM	101.2	98	92.2	Microcephaly 5, primary, autosomal recessive, 608716

ASS1	97.9	95.7	87.5	Citrullinemia, 215700
ATCAY	146.1	100	99.7	Ataxia, cerebellar, Cayman type, 601238
ATF6	134.1	100	99.6	Achromatopsia 7, 616517
ATIC	119.5	99.7	99	AICA-ribosiduria due to ATIC deficiency, 608688
ATM	109.7	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATOH7	102.8	95.8	89.6	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	117.4	100	98.8	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP2A1	155.9	100	100	Brody myopathy, 601003
ATP6V0A2	130	100	99.3	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	116.4	99.9	98.6	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	176.6	100	100	Renal tubular acidosis with deafness, 267300
ATP7B	168.9	100	99.8	Wilson disease, 277900
ATP8A2	133.5	100	99.5	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	139	96.7	94.4	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	101.4	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	138.3	99.4	96.9	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
AUH	90.9	99.9	97.6	3-methylglutaconic aciduria, type I, 250950
AURKC	79	99.8	97.5	Spermatogenic failure 5, 243060
B2M	252.1	100	99.9	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B3GALNT2	115	92.4	89.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	47.5	76.4	71.7	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	93.6	99.4	95.9	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600

B3GLCT	101.2	97.4	93.4	Peters-plus syndrome, 261540
B4GALNT1	151	95.6	90.1	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	105.4	99.9	99	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	104.3	96.1	95	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	120.4	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D2	110.9	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	121.4	98.3	95.3	Hypercholanemia, familial, 607748
BANF1	58.3	98	88.1	Nestor-Guillermo progeria syndrome, 614008
BBS1	148.9	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.6	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.6	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.8	100	99.8	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.9	99.7	97.3	Bardet-Biedl syndrome 4, 615982
BBS5	81.1	95.8	84.1	Bardet-Biedl syndrome 5, 615983
BBS7	120.7	98.1	91.7	Bardet-Biedl syndrome 7, 615984
BBS9	112.9	96	93.8	Bardet-Biedl syndrome 9, 615986
BCKDHA	171.5	100	99.5	Maple syrup urine disease, type Ia, 248600
BCKDHB	112.6	88.9	81.3	Maple syrup urine disease, type Ib, 248600
BCKDK	178.8	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCS1L	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BFSP1	98	98.2	88.9	Cataract 33, multiple types, 611391
BFSP2	89.5	99.8	97.6	Cataract 12, multiple types, 611597
BHLHA9	13.8	57.8	41.3	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BIN1	100.2	99.1	95.3	Centronuclear myopathy 2, 255200
BLM	116.3	99.4	96.5	Bloom syndrome, 210900
BLNK	95.7	93.7	91.3	?Agammaglobulinemia 4, 613502
BLOC1S3	28.7	88.7	65.3	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	97.2	98.7	91.3	?Hermansky-pudlak syndrome 9, 614171

BLVRA	125.1	100	99.7	Hyperbiliverdinemia, 614156
BMP1	143.9	99.9	99.1	Osteogenesis imperfecta, type XIII, 614856
BMPER	159.9	99.9	99	Diaphanospondylodysostosis, 608022
BMPR1B	172.4	100	98.9	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BOLA3	50.1	92.3	81.7	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	130.8	100	100	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAT1	108.5	99.8	97.4	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRF1	100	96.6	92.9	Cerebellofaciodental syndrome, 616202
BSCL2	113.5	100	100	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BSND	137.1	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	166.6	100	99.9	Biotinidase deficiency, 253260
BUB1B	136.5	98.6	97.9	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf4	131.3	99.1	94.7	Mental retardation, autosomal recessive 66, 618221
C12orf57	152	100	100	Temtamy syndrome, 218340
C12orf65	88.2	97.3	91.9	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	124.9	99.9	97.9	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	93.9	100	99.7	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1QA	120.4	100	99	C1q deficiency, 613652
C1QB	183.4	100	99.9	C1q deficiency, 613652
C1QBP	80.7	81.7	71	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	198.1	100	98.9	C1q deficiency, 613652
C1S	117.6	100	99.7	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174

C2	129.9	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf59	145.8	98.7	94.6	Ciliary dyskinesia, primary, 26, 615500
C2CD3	143.1	95.8	95.6	?Orofaciodigital syndrome XIV, 615948
C2orf71	124.6	99.7	98.8	Retinitis pigmentosa 54, 613428
C3	145.5	100	99.7	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	91.5	98.1	95.9	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	90.5	98.5	96.5	C4B deficiency, 614379
C4orf26	197.6	100	100	Amelogenesis imperfecta, type IIA4, 614832
C5	134.4	98.4	95.3	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	122.8	98.6	95.5	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8A	120.2	100	99.8	C8 deficiency, type I, 613790
C8B	135.8	99.9	99.5	C8 deficiency, type II, 613789
C8orf37	126.4	100	99	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	133.7	100	98.5	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CA12	109.6	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	124.1	99.5	94.9	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	114.6	96.8	93	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	67.1	74.6	65.5	Deafness, autosomal recessive 93, 614899
CABP4	98.4	99.7	97.7	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA2D4	112	99.2	97.7	Retinal cone dystrophy 4, 610478
CAD	158.9	100	99.7	Epileptic encephalopathy, early infantile, 50, 616457
CANT1	142.1	100	99.8	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	146	100	100	Spastic paraplegia 76, autosomal recessive, 616907

CAPN10	95.9	99.9	98	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN3	111.4	99	96.7	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600
CARD11	154.6	99.9	98.6	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD9	119.7	98.3	96.4	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	121.1	100	99.8	Combined oxidative phosphorylation deficiency 27, 616672
CASP8	144.8	95.6	95.5	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ2	143.3	99.9	99.2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	178	100	99.7	Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	110.2	96.8	92.8	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	148.5	100	100	Acatalasemia, 614097
CATSPER1	147.2	100	99.4	Spermatogenic failure 7, 612997
CAV1	265.4	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 Pulmonary hypertension, primary, 3, 615343
CAVIN1	137	99.9	99.3	Lipodystrophy, congenital generalized, type 4, 613327
CBS	116.2	97.1	91.1	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	96.6	99.9	98.4	?46XY sex reversal 5, 613080
CC2D1A	119.4	99.8	98.3	Mental retardation, autosomal recessive 3, 608443
CC2D2A	127.4	99.5	97.1	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	75.9	98.9	95.5	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	116.8	100	99	Ciliary dyskinesia, primary, 17, 614679

CCDC114	120.7	100	99.6	Ciliary dyskinesia, primary, 20, 615067
CCDC115	59.3	88	85.3	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	116.2	100	99.7	Ciliary dyskinesia, primary, 30, 616037
CCDC174	133.1	98.1	93.3	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC39	74.3	96.6	88.9	Ciliary dyskinesia, primary, 14, 613807
CCDC40	126.5	98.9	97.8	Ciliary dyskinesia, primary, 15, 613808
CCDC65	105.9	99.7	97.6	Ciliary dyskinesia, primary, 27, 615504
CCDC8	111.9	100	100	3-M syndrome 3, 614205
CCDC88A	78.9	94.7	84.9	?PEHO syndrome-like, 617507
CCDC88C	101.4	99.8	97.4	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCNO	103.1	99	95.6	Ciliary dyskinesia, primary, 29, 615872
CCT5	164.5	99.9	99.1	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	132.3	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	88.8	99.9	98.4	Immunodeficiency, common variable, 3, 613493
CD247	101.6	100	98.9	?Immunodeficiency 25, 610163
CD27	118.1	100	99.6	Lymphoproliferative syndrome 2, 615122
CD2AP	98.2	99.6	96	Glomerulosclerosis, focal segmental, 3, 607832
CD320	92	100	99.5	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	123.3	99.2	95.7	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia], 0 {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD3D	193.8	100	100	Immunodeficiency 19, 615617
CD3E	152.1	100	99.9	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	156.8	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	165.4	100	99.9	Immunodeficiency with hyper-IgM, type 3, 606843
CD59	200.9	93.6	86.5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	128.3	99.8	97	Agammaglobulinemia 3, 613501
CD79B	210.7	100	100	Agammaglobulinemia 6, 612692
CD81	142.6	99.9	98.1	Immunodeficiency, common variable, 6, 613496

CD8A	110	99.9	99	CD8 deficiency, familial, 608957
CDAN1	97.6	97.6	95.2	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	161.4	98.3	93.5	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC45	160.7	99.4	97.5	Meier-Gorlin syndrome 7, 617063
CDC6	165.4	99.8	98.3	?Meier-Gorlin syndrome 5, 613805
CDCA7	109.2	100	99.3	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH23	197.2	100	100	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CDH3	159.3	99.5	97.3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	154.2	99.2	98	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	114.6	100	99.9	Al Kaissi syndrome, 617694
CDK5RAP2	123.9	99.9	98.7	Microcephaly 3, primary, autosomal recessive, 604804
CDSN	119.3	100	99.5	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99.3	96.8	93.8	Meier-Gorlin syndrome 4, 613804
CEBPE	71.1	99.3	95.8	Specific granule deficiency, 245480
CENPF	139.5	99.5	97.6	Stromme syndrome, 243605
CENPJ	141.7	99.7	97.8	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	119.9	99	97.9	Joubert syndrome 25, 616781
CEP120	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	79.2	98.1	89.1	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	162.5	97.2	94.5	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	94.2	99.9	98	Nephronophthisis 15, 614845
CEP19	202.7	100	100	Morbid obesity and spermatogenic failure, 615703
CEP290	66.1	88.4	76.7	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755

				Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	83.5	97.7	89.6	Joubert syndrome 15, 614464
CEP57	92.6	99.4	93.3	Mosaic variegated aneuploidy syndrome 2, 614114
CEP83	96.7	98.3	89.1	Nephronophthisis 18, 615862
CEP89	125.5	94.7	91.4	No OMIM phenotype Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138) ?Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679)
CERKL	100.4	98.6	92.8	Retinitis pigmentosa 26, 608380
CERS3	106.8	100	98.8	Ichthyosis, congenital, autosomal recessive 9, 615023
CFAP53	146.6	97.6	94.2	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFD	80.6	89.7	81.6	Complement factor D deficiency, 613912
CFH	183.2	98.7	95.3	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFI	145.5	96.6	92.8	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	117.3	94.3	86.7	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	124	99.1	96.3	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800
CHAT	130.3	89.3	86.8	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	133.7	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHRNA1	121.8	94.7	94.6	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	131.8	98.8	96.7	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313

CHRND	150.5	100	99	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	127.7	99.3	95.8	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNG	155.2	100	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	165.6	95.7	93.3	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	91.6	100	97.5	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	334	100	100	Macular corneal dystrophy, 217800
CHSY1	138.4	95.9	93.9	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	131.6	100	98.4	Cocoon syndrome, 613630
CIB2	229.9	99.9	99.6	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIITA	125	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CISD2	127.9	83.4	83.4	Wolfram syndrome 2, 604928
CIT	108.6	99.9	98.2	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	161.3	98.9	96.6	Filippi syndrome, 272440
CLCF1	76.2	98.7	97.7	Cold-induced sweating syndrome 2, 610313
CLCN1	137.4	100	99.5	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	108.6	100	99.4	Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN7	129.7	99.5	98.2	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLCNKB	100.7	98.5	90.5	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090

CLDN1	137.6	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN14	130.6	100	99.9	Deafness, autosomal recessive 29, 614035
CLDN16	136.3	100	99.9	Hypomagnesemia 3, renal, 248250
CLDN19	123.7	98.2	93.7	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLIP1	131.7	99.7	97.8	No OMIM phenotype Intellectual disability, autosomal recessive (Larti (2015) Eur J Hum Genet 23,331)
CLMP	111	100	99.9	Congenital short bowel syndrome, 615237
CLN3	114.9	92.5	90.7	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.1	98.2	92.2	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.6	98.9	95.3	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	163.9	83.5	83.5	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	182.4	100	99.8	Pontocerebellar hypoplasia, type 10, 615803
CLPB	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	115.4	99.8	96.9	Perrault syndrome 3, 614129
CLRN1	157.2	100	99.8	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNGA1	127.2	89.4	84.6	Retinitis pigmentosa 49, 613756
CNGA3	167.7	100	99.9	Achromatopsia 2, 216900
CNGB1	102.5	98.4	94.8	Retinitis pigmentosa 45, 613767
CNGB3	101.4	97.7	93	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200
CNNM2	188.4	100	99.2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	190.5	98.7	97.8	Jalili syndrome, 217080
CNPY3	87.1	99.8	96.7	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	151.7	99.8	98.3	?Myopathy, congenital, Compton-North, 612540
CNTNAP1	161.2	99.2	97.5	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286
CNTNAP2	148	100	99.9	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA6	78.7	98.8	91.9	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501

COASY	168.5	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COG1	124.2	100	99.9	Congenital disorder of glycosylation, type IIg, 611209
COG4	123.8	100	99.9	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	107	97.4	93.8	Congenital disorder of glycosylation, type IIi, 613612
COG6	78.4	95	85.9	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	125.1	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	122.4	99.9	98.4	Congenital disorder of glycosylation, type IIh, 611182
COL11A1	90.8	94.9	89.6	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	92.2	99.9	98.3	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL12A1	137.5	99.5	97.5	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL13A1	85.4	99.8	97.1	Myasthenic syndrome, congenital, 19, 616720
COL17A1	107.9	99.2	96.6	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	88.7	93.9	87.7	Knobloch syndrome, type 1, 267750
COL1A2	101.8	96.7	93.6	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	126.8	98.4	95.8	Fibrosis of extraocular muscles, congenital, 5, 616219
COL4A3	89.6	97.8	95.5	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200

COL4A4	85	97.6	93.5	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign, 0
COL6A1	137.2	99.5	97.8	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	165.3	99.3	98.4	?Myosclerosis, congenital, 255600 Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	174.7	100	99.9	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	129.5	99.6	97.5	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant, 0 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COL9A1	121.2	99.5	96.9	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	65.1	98.3	88.8	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COLEC11	203	100	100	3MC syndrome 2, 265050
COLQ	113.2	99.8	98.1	Myasthenic syndrome, congenital, 5, 603034
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88.4	84.9	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	143.9	99.3	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ8A	134.3	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	90.5	100	99.1	Nephrotic syndrome, type 9, 615573
COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
CORO1A	154.4	99.8	96.9	Immunodeficiency 8, 615401
COX10	241.9	100	99.6	Leigh syndrome due to mitochondrial COX4 deficiency, 256000

				Mitochondrial complex IV deficiency, 220110
COX15	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	58.1	83	65.4	Mitochondrial complex IV deficiency, 220110
COX4I2	120.1	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	180.6	100	99.4	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	159.6	100	100	Mitochondrial complex IV deficiency, 220110
CP	120	93.9	89.6	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	118.3	99.8	98.5	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPN1	118.3	100	98.8	Carboxypeptidase N deficiency, 212070
CPOX	116.8	95.2	88.1	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	143.8	100	99.8	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	169.3	100	98.7	CPT deficiency, hepatic, type IA, 255120
CPT2	162.8	97.2	95.4	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	160.7	100	99.8	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	115.2	99.9	98.5	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRB1	191.8	100	100	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105
CRB2	112.4	99.4	94.7	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	130.7	87.8	83.8	Mental retardation, autosomal recessive 2, 607417
CRIPT	34.5	96.5	74.8	Short stature with microcephaly and distinctive facies, 615789
CRLF1	105.6	90.9	89.2	Cold-induced sweating syndrome 1, 272430

CRTAP	110.4	99.8	97.3	Osteogenesis imperfecta, type VII, 610682
CRYAA	135.3	92.7	86.2	Cataract 9, multiple types, 604219
CRYBB1	129.1	100	99.4	Cataract 17, multiple types, 611544
CRYBB3	144.3	100	100	Cataract 22, 609741
CSF2RB	94.8	99.6	97.8	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSPP1	112	99.8	97.8	Joubert syndrome 21, 615636
CSTA	119	99.9	99	Peeling skin syndrome 4, 607936
CSTB	82.5	97.1	82.7	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	105	86.6	83.6	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNS	120.1	100	99.9	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	143.1	100	99.6	Immunodeficiency 24, 615897
CTSA	134.1	100	99.4	Galactosialidosis, 256540
CTSC	127.5	100	100	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	163.7	98	95.3	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	112.8	84.2	80.2	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	105	100	99.9	Pycnodysostosis, 265800
CUBN	127.8	99.8	98.4	Megaloblastic anemia-1, Finnish type, 261100
CUL7	149.6	99.8	97.9	3-M syndrome 1, 273750
CWF19L1	119.1	99.5	96.7	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5A	133.5	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	147.3	98	98	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	97.3	77.9	71	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYC1	184.5	88.1	86.8	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYP11A1	123.9	99.6	97.7	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	175.9	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	173.4	100	100	Aldosterone to renin ratio raised, 0

				Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}, 0
CYP17A1	135	100	99.7	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	160.6	99.1	97.3	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	134.8	100	100	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP24A1	165.3	100	100	Hypercalcemia, infantile, 1, 143880
CYP26B1	178.1	100	99.9	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	87.2	99.5	95.6	Focal facial dermal dysplasia 4, 614974
CYP27A1	175.1	98.3	96.1	Cerebrotendinous xanthomatosis, 213700
CYP27B1	137.1	100	99.1	Vitamin D-dependent rickets, type I, 264700
CYP2C8	109.1	98	93.7	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2R1	138.5	96.4	86.6	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	119.2	93.7	91.2	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	127.7	100	99.7	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	147.5	99.8	98.5	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	93.2	94.7	87.7	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	134.5	97.5	95.2	D-2-hydroxyglutaric aciduria, 600721
DAG1	220.8	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	98.6	98.7	93.8	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	122.3	100	99.6	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	140.7	100	99.8	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	102.1	97.3	93.8	Maple syrup urine disease, type II, 248600
DCAF17	91.9	95.6	89.3	Woodhouse-Sakati syndrome, 241080
DCC	138.5	100	99.9	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCDC2	150.5	99.9	99.6	?Deafness, autosomal recessive 66, 610212

				Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DCHS1	160.1	99.8	99.2	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	128.8	98.2	94.5	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCPS	143.8	100	99.8	Al-Raqad syndrome, 616459
DDB2	162.4	100	99.7	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	101	99.1	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	141.8	97.1	94.8	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	149.7	99.9	98	Spastic paraplegia 54, autosomal recessive, 615033
DDR2	155	100	99.9	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDX11	113.9	86	81	Warsaw breakage syndrome, 613398
DDX59	151.7	99.7	97.6	Orofaciodigital syndrome V, 174300
DENND5A	123	99.8	97.9	Epileptic encephalopathy, early infantile, 49, 617281
DFNB59	123.6	100	99.2	Deafness, autosomal recessive 59, 610220
DGKE	142.3	99.5	95.2	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	183	100	100	Desmosterolosis, 602398
DHCR7	158.3	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	93.5	97.8	94.8	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHFR	48.4	91.1	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	117.7	100	100	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	92.2	100	99.9	Miller syndrome, 263750
DHTKD1	141	99.6	98.2	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIAPH1	120.7	99.3	97.8	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632

DIS3L2	158.5	99.8	99	Perlman syndrome, 267000
DLAT	91.6	99.1	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	123.5	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DLL3	64.1	88.8	79.9	Spondylocostal dysostosis 1, autosomal recessive, 277300
DMGDH	157	98.8	97.2	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	159.5	99.9	99.1	Hypophosphatemic rickets, AR, 241520
DNAAF1	115.8	100	99.7	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	105.1	99.7	96.9	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	91.8	97.7	90.6	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	79.6	96.3	84.1	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	107.9	84.5	78.2	Ciliary dyskinesia, primary, 18, 614874
DNAH11	134	99.8	98.4	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	123.8	99.7	98.5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	135.3	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	156.6	98.4	95.5	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	102	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJC12	129.3	87.4	87.3	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	97.9	98.5	90	3-methylglutaconic aciduria, type V, 610198
DNAJC21	125.7	99.8	98.5	Bone marrow failure syndrome 3, 617052
DNAJC6	160.8	99.9	98.9	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	99	95.7	84.5	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	141.3	100	100	Systemic lupus erythematosus 16, 614420
DNM1L	123.5	99.7	96.6	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNMT3B	124.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	143.8	100	99.9	Immunodeficiency 40, 616433
DOCK6	119.9	98.9	96.5	Adams-Oliver syndrome 2, 614219
DOCK7	114.4	97.9	95.6	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	129.1	100	99.8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	105.7	93.3	92.5	?Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300
DOLK	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768

DONSON	104.9	83.9	78.3	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	157.1	100	99.7	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.1	100	99.4	Congenital disorder of glycosylation, type Iu, 615042
DPM3	183.9	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPY19L2	94.8	72.9	66.6	Spermatogenic failure 9, 613958
DPYD	158.3	95.6	93.7	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	133.5	100	99.5	Dihydropyrimidinuria, 222748
DRAM2	131.8	100	99.7	Cone-rod dystrophy 21, 616502
DRC1	97	99.9	98.6	Ciliary dyskinesia, primary, 21, 615294
DSC2	128.5	99.4	96.2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	87.1	97.2	89.6	?Hypotrichosis and recurrent skin vesicles, 613102
DSG4	198.4	98.5	95.7	Hypotrichosis 6, 607903
DSP	154	100	99.8	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DST	154.1	99.7	98.2	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DTNBP1	115.2	99.3	95.1	Hermansky-Pudlak syndrome 7, 614076
DUOX2	136.4	96.7	94.8	Thyroid dyshormonogenesis 6, 607200
DUOXA2	122.2	100	99.9	Thyroid dyshormonogenesis 5, 274900
DYM	101.3	97.2	94.8	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC2H1	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95.1	99.3	96	Short-rib thoracic dysplasia 15 with polydactyly, 617088

DYSF	133.1	100	99.8	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
EARS2	103.4	99.7	98.3	Combined oxidative phosphorylation deficiency 12, 614924
ECEL1	100.7	88.8	83.1	Arthrogryposis, distal, type 5D, 615065
ECHS1	112.8	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	170.8	100	99.7	Urbach-Wiethe disease, 247100
EDAR	138.6	100	99.6	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	99.1	99.3	93.3	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDC3	144.7	100	99.6	?Mental retardation, autosomal recessive 50, 616460
EDN1	145.5	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDN3	134.4	100	99.5	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	131	95.6	90.9	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EFEMP2	120.9	100	99.9	Cutis laxa, autosomal recessive, type IB, 614437
EGF	135.2	100	99.8	Hypomagnesemia 4, renal, 611718
EGFR	160.8	100	99.1	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGR2	124.4	100	100	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
EIF2AK3	147.1	95.1	91.3	Wolcott-Rallison syndrome, 226980
EIF2AK4	146.3	99.8	98	Pulmonary venoocclusive disease 2, 234810
EIF2B1	149.9	100	100	Leukoencephalopathy with vanishing white matter, 603896

EIF2B2	131.9	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	163.8	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	146	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	128	99.6	97.9	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	106.8	100	99.9	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	123.8	100	99.3	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELOVL4	91.9	99.9	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELP1	142.6	99.7	98.1	Dysautonomia, familial, 223900
ELP2	125.5	99.2	96.9	Mental retardation, autosomal recessive 58, 617270
EMC1	124.3	100	99.8	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMG1	137.7	100	100	Bowen-Conradi syndrome, 211180
EMP2	101.1	99.7	96.9	Nephrotic syndrome, type 10, 615861
ENAM	148.9	100	99.9	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENO3	179.3	100	100	?Glycogen storage disease XIII, 612932
ENPP1	134.8	92.4	83.2	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
ENTPD1	165.1	100	99.4	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	119.9	79.5	78.4	Adams-Oliver syndrome 4, 615297
EPB41	135.5	98.7	95.6	Elliptocytosis-1, 611804
EPB42	160.7	100	99.7	Spherocytosis, type 5, 612690
EPCAM	64.5	93.3	79.8	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	126	99.3	97.7	Vici syndrome, 242840
EPHX1	122.6	98.8	96.1	?Hypercholanemia, familial, 607748

EPM2A	110.1	86.2	84	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPRS	126.3	100	99.2	Leukodystrophy, hypomyelinating, 15, 617951
ERBB3	139.2	100	99.9	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
ERCC1	76.6	100	97.3	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	123.7	100	99.7	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.2	99.9	98.9	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	139.2	100	99.5	?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	139.8	100	99.4	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	191.3	100	99.9	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC6L2	107.6	99.7	97.7	Bone marrow failure syndrome 2, 615715
ERCC8	89.5	92.9	78.4	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN1	146.9	100	100	Spastic paraplegia 62, 615681
ERLIN2	156	100	99.3	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	105.2	97.3	90.4	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	28.6	44.2	35.3	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 0
ESR1	127.5	99.9	99.4	Estrogen resistance, 615363 {Atherosclerosis, susceptibility to}, 0

				{Breast cancer}, 114480 {HDL response to hormone replacement, augmented}, 0 {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESRRB	119.7	100	99.2	Deafness, autosomal recessive 35, 608565
ETFA	143.3	100	99.4	Glutaric acidemia IIA, 231680
ETFB	126.6	100	100	Glutaric acidemia IIB, 231680
ETFDH	105.4	100	99.3	Glutaric acidemia IIC, 231680
ETHE1	85.5	99.3	95.8	Ethylmalonic encephalopathy, 602473
EVC	110.4	93.2	89.8	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	119.3	96.4	94.3	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOSC2	142	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	88.5	97.3	89.4	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	80.1	91.8	76.9	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	129.8	97.5	87.4	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	183.3	100	99.9	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EYS	135.8	98.9	94.9	Retinitis pigmentosa 25, 602772
F10	185.1	99	98.3	Factor X deficiency, 227600
F11	155	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	111.4	100	99.5	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	147.3	100	99.4	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	113.5	96.6	87.6	Factor XIIIB deficiency, 613235
F2	124.2	99.8	98.1	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	173.5	99	97.3	Factor V deficiency, 227400

				Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	166.6	100	98.5	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
FA2H	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319
FADD	142.8	100	99.6	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	151.3	100	99.9	Tyrosinemia, type I, 276700
FAM126A	125.2	97.3	95.2	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	115.2	98.5	95	Retinitis pigmentosa 28, 606068
FAM20A	105.4	98.4	92.1	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	101.3	100	98.9	Raine syndrome, 259775
FANCA	123.3	99.8	98.5	Fanconi anemia, complementation group A, 227650
FANCB	68.4	96.7	87.9	Fanconi anemia, complementation group B, 300514
FANCC	121.6	99.4	97.1	Fanconi anemia, complementation group C, 227645
FANCD2	127.6	98.7	95.5	Fanconi anemia, complementation group D2, 227646
FANCE	108	85.9	84.6	Fanconi anemia, complementation group E, 600901
FANCF	166.8	100	100	Fanconi anemia, complementation group F, 603467
FANCG	147.7	100	100	Fanconi anemia, complementation group G, 614082
FANCI	152.1	99.5	97.5	Fanconi anemia, complementation group I, 609053
FANCL	87.8	99.4	94.7	Fanconi anemia, complementation group L, 614083
FAR1	80.4	96.3	92.4	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	207.7	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FASTKD2	118.9	99.5	96.8	?Mitochondrial complex IV deficiency, 220110
FAT4	224.5	100	99.9	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	119.6	91.8	91.1	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895

FBP1	127	100	98.8	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	189.8	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	108.8	93.5	89	?Mental retardation, autosomal recessive 45, 615979
FBXO7	189.4	98.5	93.3	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	225	99.6	97.7	Immunodeficiency 20, 615707
FCN3	127.8	100	99.4	Immunodeficiency due to ficolin 3 deficiency, 613860
FDXR	93.4	100	99.1	Auditory neuropathy and optic atrophy, 617717
FECH	121.9	99.9	99.4	Protoporphyria, erythropoietic, 1, 177000
FERMT1	104.9	98.9	96.3	Kindler syndrome, 173650
FERMT3	122.4	100	98.9	Leukocyte adhesion deficiency, type III, 612840
FEZF1	158.8	99.9	99.3	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	157	99	96.6	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	190.8	99.7	97.9	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD4	111.9	99.3	97.3	Charcot-Marie-Tooth disease, type 4H, 609311
FGF23	106	99.9	97.8	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF3	73.9	92	75.7	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGG	137	99.3	96.5	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	123.3	100	100	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	154.9	99.8	98.4	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340

FKBP10	158.6	96.9	92.8	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	74.3	100	99.4	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	94.5	100	99.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	120	99.2	94.2	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	191.6	100	98.9	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLG	234.1	100	99.9	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNB	149.9	99.8	99.2	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLVCR1	139.5	99.2	95.8	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	159.7	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	86.6	83.1	77.7	Mental retardation, autosomal recessive 47, 616193
FMO3	153.5	99.9	99.2	Trimethylaminuria, 602079
FOLR1	150.4	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXE1	29.3	72.3	56.2	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXI1	152.5	100	100	Enlarged vestibular aqueduct, 600791
FOXN1	112.5	100	99.5	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXRED1	136.6	100	99.6	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	147.8	100	99.7	Fraser syndrome 1, 219000
FREM1	138.4	99.9	99.1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	182.4	100	99.5	Fraser syndrome 2, 617666

FRMD4A	116.7	91.4	90.3	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRRS1L	103.1	68.3	63.4	Epileptic encephalopathy, early infantile, 37, 616981
FSHB	149.1	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	140.9	100	98.8	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FTCD	89.8	94.6	89.8	Glutamate formiminotransferase deficiency, 229100
FTO	118.9	83.7	82.5	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FUCA1	135	100	99.5	Fucosidosis, 230000
FUT8	166.1	99.9	98.8	Congenital disorder of glycosylation with defective fucosylation, 618005
FXN	75.2	85.7	75.9	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FYCO1	123.7	100	100	Cataract 18, autosomal recessive, 610019
FZD6	208.6	100	100	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	180.7	100	100	Glycogen storage disease Ia, 232200
G6PC3	123.7	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GAA	128.5	100	99.9	Glycogen storage disease II, 232300
GAD1	128.7	99.9	98.4	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	100.6	98.9	94.6	Krabbe disease, 245200
GALE	154.8	100	100	Galactose epimerase deficiency, 230350
GALK1	125.4	100	99.7	Galactokinase deficiency with cataracts, 230200
GALNS	93.2	99	95.6	Mucopolysaccharidosis IVA, 253000
GALNT3	128.2	99.2	96	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	168.7	100	100	Galactosemia, 230400
GAMT	93.5	90.9	80.7	Cerebral creatine deficiency syndrome 2, 612736
GAN	190	100	99.9	Giant axonal neuropathy-1, 256850
GAS8	150.7	99.8	99.4	Ciliary dyskinesia, primary, 33, 616726
GATM	150.6	100	100	Cerebral creatine deficiency syndrome 3, 612718
GBA	240.3	100	100	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000

				Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	176.2	99.9	99.3	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	147.6	99.9	99.1	Glutaricaciduria, type I, 231670
GCH1	74.4	97	86.5	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	141.4	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	133.2	99.9	98.9	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCNT2	166.8	99.5	99.5	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	34.2	83.1	67.8	?Glycine encephalopathy, 605899
GDAP1	163.1	99.3	96.1	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400
GDF1	19.5	65	48.4	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF5	141.8	100	100	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GFER	76.1	92.9	75.4	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay,

				613076
GFM1	100.3	99.2	95.3	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	144.4	99.9	97.6	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	115.3	100	99.7	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GGT1	12.9	19.8	17.2	?Glutathioninuria, 231950
GH1	175.2	100	100	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	212.3	99.8	99.5	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	116.4	95.3	94.7	Growth hormone deficiency, isolated, type IV, 618157
GHSR	206	99.9	98.4	Growth hormone deficiency, isolated partial, 615925
GIF	141.2	100	99.9	Intrinsic factor deficiency, 261000
GIPC3	103.3	91.8	85.5	Deafness, autosomal recessive 15, 601869
GJA1	246.4	100	100	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB2	205.1	100	100	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB6	185.4	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645

				Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	41.9	68.9	58.6	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	78.9	90.6	82.7	Glycine encephalopathy, 605899
GLE1	110.8	100	99.7	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLIS2	109	99.9	98.2	Nephronophthisis 7, 611498
GLIS3	133.4	99.9	99.3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA1	123.3	100	100	Hyperekplexia 1, 149400
GLRB	98.6	96.6	88.8	Hyperekplexia 2, 614619
GLRX5	108.2	92.6	83.8	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUL	108.8	99.9	98.2	Glutamine deficiency, congenital, 610015
GLYCTK	202.6	100	99.5	D-glyceric aciduria, 220120
GM2A	139.6	100	100	GM2-gangliosidosis, AB variant, 272750
GMPPA	136.8	100	99.9	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	228.5	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAT2	130.7	99.9	99.1	Achromatopsia 4, 613856
GNB3	179	100	100	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB5	125.9	99.9	98.3	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	153.7	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	147.2	99.4	97	Glycine N-methyltransferase deficiency, 606664
GNPAT	133.6	99.4	96.4	Rhizomelic chondrodysplasia punctata, type 2, 222765

GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	151.6	96.1	89.7	Mucolipidosis III gamma, 252605
GNRHR	161.9	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	107.9	96.9	92	Mucopolysaccharidosis type IIID, 252940
GORAB	176.3	99.7	97.8	Geroderma osteodysplasticum, 231070
GOSR2	127.2	95.9	95	Epilepsy, progressive myoclonic 6, 614018
GP1BA	153	97	94.3	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	34.5	74.2	64.3	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	136.5	100	100	Bleeding disorder, platelet-type, 11, 614201
GP9	123.3	96.6	89.3	Bernard-Soulier syndrome, type C, 231200
GPAA1	130	96.4	95.2	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC6	142	100	100	Omodysplasia 1, 258315
GPD1	93.5	99.9	99.1	Hypertriglyceridemia, transient infantile, 614480
GPHN	167.2	98.4	96.9	Molybdenum cofactor deficiency C, 615501
GPI	142.6	100	99.5	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	119.7	99.9	99.2	Hyperlipoproteinemia, type 1D, 615947
GPNMB	177.8	100	100	Amyloidosis, primary localized cutaneous, 3, 617920
GPR179	133.6	100	99.7	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPSM2	112.9	99.8	97	Chudley-McCullough syndrome, 604213
GPT2	138	98.2	90.9	Mental retardation, autosomal recessive 49, 616281
GPX4	119.2	85.2	76.6	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRHPR	112.5	85.1	78.2	Hyperoxaluria, primary, type II, 260000
GRID2	175.4	100	99.9	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	133.2	96	94.7	Mental retardation, autosomal recessive, 6, 611092
GRIP1	130.8	100	99.9	Fraser syndrome 3, 617667
GRK1	126.4	100	99.9	Oguchi disease-2, 613411
GRM1	185.8	100	99.9	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	151.7	93.3	86.7	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270

GRN	184.5	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	183.7	100	99.8	Deafness, autosomal recessive 25, 613285
GSC	85.4	86.9	74.5	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSS	104	100	99.8	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	83.5	96.5	91	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	113.6	100	99.1	Trichothiodystrophy 3, photosensitive, 616395
GTPBP3	137.4	100	99.7	Combined oxidative phosphorylation deficiency 23, 616198
GUCY1A3	171.3	99.4	98	Moyamoya 6 with achalasia, 615750
GUCY2C	135.2	100	99.7	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	91.3	98.3	91.1	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUSB	116.1	92.2	89.4	Mucopolysaccharidosis VII, 253220
GYG1	157.8	100	99.6	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	110.8	100	98.5	Glycogen storage disease 0, muscle, 611556
GYS2	150.2	98.5	93.9	Glycogen storage disease 0, liver, 240600
H6PD	169.5	99	99	Cortisone reductase deficiency 1, 604931
HACE1	125.3	99.2	95	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	110.8	98	95.1	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	84.4	96.5	90.3	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.5	92.5	79.5	Trifunctional protein deficiency, 609015
HAMP	175.6	100	100	Hemochromatosis, type 2B, 613313
HARS	159.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738

HBB	176.7	100	100	Delta-beta thalassemia, 141749 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methmoglobinemia, beta type, 617971 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Thalassemia-beta, dominant inclusion-body, 603902 {Malaria, resistance to}, 611162
HELLS	93.7	94.1	86.8	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	142.3	81.4	76.1	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	173.7	99.9	99.4	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	114.4	80.9	77.9	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	29.1	64.9	42.6	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	57.6	99.2	92.6	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	118.3	93.8	92.2	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	129.7	99.4	94	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	142	100	99.7	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	116.8	100	100	Hemochromatosis, type 2A, 602390
HFM1	42.8	90.3	76.7	Premature ovarian failure 9, 615724
HGD	127.8	100	99.8	Alkaptonuria, 203500
HGF	146.9	99.4	96.9	Deafness, autosomal recessive 39, 608265

HGSNAT	101	86.4	85.7	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	67.7	92.7	69.5	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	61.7	90.9	79.7	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	60	98.5	88	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	143.4	100	99.9	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HLCS	172.8	100	100	Holocarboxylase synthetase deficiency, 253270
HMGCL	143.3	100	99.9	HMG-CoA lyase deficiency, 246450
HMGCS2	131.5	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	128.7	95.8	89.5	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	23	56.8	42	Oculoauricular syndrome, 612109
HNMT	135	100	99.7	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HOGA1	147.5	99.8	98.1	Hyperoxaluria, primary, type III, 613616
HOXA1	165.3	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA2	74.1	99.5	95.6	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HOXB1	104.5	100	100	Facial paresis, hereditary congenital, 3, 614744
HOXC13	104.9	97.5	91.1	Ectodermal dysplasia 9, hair/nail type, 614931
HPCA	283.4	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	137.8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	88	100	98.5	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPS1	117.8	100	99.3	Hermansky-Pudlak syndrome 1, 203300
HPS3	135.2	99.6	96.4	Hermansky-Pudlak syndrome 3, 614072
HPS4	141.9	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	133	99.9	98.7	Hermansky-Pudlak syndrome 5, 614074
HPS6	139.1	91	84.3	Hermansky-Pudlak syndrome 6, 614075

HPSE2	110.2	98.8	95.4	Urofacial syndrome 1, 236730
HR	94.9	97.3	94.2	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HSD11B2	165.2	85.7	82.5	Apparent mineralocorticoid excess, 218030
HSD17B3	156.4	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95.1	93.9	90.8	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	189.2	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	136.3	98.2	91	Bile acid synthesis defect, congenital, 1, 607765
HSPA9	91.6	91.1	85.9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	96.5	98.3	93.2	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	121.3	99.4	98.2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTRA1	98.2	84.5	81.4	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149
HYAL1	115.3	100	100	?Mucopolysaccharidosis type IX, 601492
HYDIN	133.6	99.9	99.5	Ciliary dyskinesia, primary, 5, 608647
HYLS1	171.1	100	100	Hydrolethalus syndrome, 236680
IARS	148.8	99.8	98.6	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093
IARS2	131.5	100	99.9	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ICK	131.6	99.9	98.2	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
ICOS	160.2	100	100	Immunodeficiency, common variable, 1, 607594
IDH3B	165.5	95.9	95.4	Retinitis pigmentosa 46, 612572
IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016

IER3IP1	73	93.2	82.2	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFNGR1	138.5	99.2	97.3	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	142.3	93.2	93.1	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT140	114.7	99.9	99	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT80	57.8	87.6	70.7	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92.9	88.3	81.2	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	122.5	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	144	100	99.8	Insulin-like growth factor I, resistance to, 270450
IGFALS	79.7	99.9	96.8	Acid-labile subunit, deficiency of, 615961
IGFBP7	71.6	91.7	83.5	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHM	185.2	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	107.8	99.3	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	157.9	100	100	Kappa light chain deficiency, 614102
IGLL1	86.2	99.3	94.9	Agammaglobulinemia 2, 613500
IHH	129.3	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	123.5	98.5	94.2	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IL10RA	141.9	100	99.9	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	168.8	98.7	96.1	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424

IL11RA	139.9	100	99.5	Craniosynostosis and dental anomalies, 614188
IL12B	121.1	100	99.9	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	124.3	97	94.7	Immunodeficiency 30, 614891
IL17RC	96.1	99.8	99	Candidiasis, familial, 9, 616445
IL1RN	162.8	100	100	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL21R	128.5	100	100	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050
IL2RA	116.4	100	99.5	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL36RN	99	100	100	Psoriasis 14, pustular, 614204
IL7R	129.5	99.9	99.4	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	107.4	100	99.9	Deafness, autosomal recessive 42, 609646
IMPA1	71.4	97.1	85.8	Mental retardation, autosomal recessive 59, 617323
IMPAD1	147.2	99.9	99.4	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPG2	154.3	99.5	97.8	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	108.3	100	99.6	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	123.6	96.7	93.7	Opsismodysplasia, 258480
INSR	141.1	97.1	94.5	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INVS	160.5	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89.3	75.4	Senior-Loken syndrome 5, 609254
IRAK4	95.4	98.3	90.1	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF8	114.7	99.6	97.4	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRX5	74.7	94.2	86.1	Hamamy syndrome, 611174
ISCA2	92	99.7	96.9	Multiple mitochondrial dysfunctions syndrome 4, 616370

ISCU	111.2	100	99.7	Myopathy with lactic acidosis, hereditary, 255125
ISG15	160.1	100	100	Immunodeficiency 38, 616126
ISPD	104.4	95.2	84.8	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	124.7	95.4	94.8	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	107.2	99.6	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	141.5	99.8	98.3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	146.5	99.8	99	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	129.6	99.6	97.6	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	122.4	99.6	98.1	Renal hypodysplasia/aplasia 1, 191830
ITGB2	152.1	100	99.8	Leukocyte adhesion deficiency, 116920
ITGB3	142.2	99.3	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
ITGB4	150.1	97.4	94.8	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	137	96.5	95.2	Amelogenesis imperfecta, type IH, 616221
ITK	125.2	100	99.6	Lymphoproliferative syndrome 1, 613011
ITPA	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
IVD	114.9	100	100	Isovaleric acidemia, 243500
IYD	117.3	99.7	97.8	Thyroid dyshormonogenesis 4, 274800
JAGN1	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK3	104.2	98.2	95.2	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	158.6	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JUP	145.1	100	99.6	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KALRN	149	100	99.5	{Coronary heart disease, susceptibility to, 5}, 608901
KANK2	151.4	99.9	99.5	Nephrotic syndrome, type 16, 617783

				Palmoplantar keratoderma and woolly hair, 616099
KARS	122.6	100	99.3	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KATNB1	141.7	100	100	Lissencephaly 6, with microcephaly, 616212
KCNE1	462.6	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNJ1	233.9	100	100	Bartter syndrome, type 2, 241200
KCNJ10	213.4	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	299.5	100	100	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	210.4	100	99.9	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNQ1	114.7	93	90.3	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNV2	137.7	100	100	Retinal cone dystrophy 3B, 610356
KCTD7	166.7	95	95	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KERA	191.6	100	100	Cornea plana 2, autosomal recessive, 217300
KHDC3L	110.3	99.9	99.2	Hydatidiform mole, recurrent, 2, 614293
KIAA0586	114.7	98.2	92.7	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1109	144.1	99.1	97.2	Alkuraya-Kucinskas syndrome, 617822
KIF1A	114	99.2	96.1	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1BP	159.7	96.2	96.1	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	121.3	99.9	99.1	Spastic ataxia 2, autosomal recessive, 611302
KIF7	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131

				?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KISS1R	106.4	99.5	95.3	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
KIZ	174.2	98.3	96	Retinitis pigmentosa 69, 615780
KL	179.9	97.2	96	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	115.9	99.8	98.2	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLHL3	141.1	100	99.8	Pseudohypoaldosteronism, type IID, 614495
KLHL40	157.9	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	203.5	100	99.6	Nemaline myopathy 9, 615731
KLK4	185.1	100	98.8	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	143.4	99.6	96.7	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	120.3	94	91.2	Dystonia 28, childhood-onset, 617284
KNL1	113.7	98.3	95.2	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	112.1	100	99.9	Mental retardation, autosomal recessive 41, 615637
KRT10	103	98.7	93.9	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT14	59.3	89	82.1	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000
KRT18	40.2	84.8	67.8	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT8	39.2	91.7	73.4	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT85	108.3	98.8	95.3	Ectodermal dysplasia 4, hair/nail type, 602032
L2HGDH	129.1	98.4	97	L-2-hydroxyglutaric aciduria, 236792
LAMA1	137.5	100	99.6	Poretti-Boltshauser syndrome, 615960
LAMA2	143.5	99.9	99.5	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138

LAMA3	147.6	99.7	99.2	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMB1	169.8	100	99.6	Lissencephaly 5, 615191
LAMB2	201.7	100	99.8	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	123.4	100	99.6	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	117.6	99.9	98.8	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	130.5	98.3	96.1	Cortical malformations, occipital, 614115
LAMTOR2	167	100	99.9	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	143	100	99.6	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	56.3	80.5	63.7	Alazami syndrome, 615071
LARS2	143	100	100	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300
LBR	87.8	93.3	83.9	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	127.8	97.3	95.7	Leber congenital amaurosis 5, 604537
LCAT	145.6	97.8	91.7	Fish-eye disease, 136120 Norum disease, 245900
LCT	142	99.9	99.1	Lactase deficiency, congenital, 223000
LDHA	59.8	94.1	87.1	Glycogen storage disease XI, 612933
LDLRAP1	156.1	95.7	91.2	Hypercholesterolemia, familial, autosomal recessive, 603813
LEMD2	68.6	89.1	80.6	Cataract 46, juvenile-onset, 212500
LEP	188.8	100	99.6	Obesity, morbid, due to leptin deficiency, 614962
LEPR	109.6	93.8	90.2	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	29	97.2	73.2	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	154.5	95.4	92.8	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320

				Leydig cell hypoplasia with pseudohermaphroditism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	287.7	100	100	Deafness, autosomal recessive 67, 610265
LHX3	84.7	94	80.6	Pituitary hormone deficiency, combined, 3, 221750
LIAS	133.7	99.5	97.1	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	123.4	97.2	92.1	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	103.2	100	98.6	Cataract 19, multiple types, 615277
LIMS2	110.8	93	92.3	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINS1	147.8	99.9	98	Mental retardation, autosomal recessive 27, 614340
LIPA	110.9	98.8	95.8	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	115.2	100	99.8	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	113.4	99.9	98.4	Lipodystrophy, familial partial, type 6, 615980
LIPH	140.6	100	100	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	125.2	99.2	95.8	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	227.4	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	92.2	97.3	83.2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN1	129.8	99.3	94.4	Combined factor V and VIII deficiency, 227300
LMAN2L	127.9	100	99.8	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	94	95.9	88.1	Acheiropody, 200500 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750 Polydactyly, preaxial type II, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	80.2	91.9	83	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	132.4	99.6	97.8	Lipase deficiency, combined, 246650

LMNA	89.2	97.9	91.3	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMOD3	141.5	99.9	98.5	Nemaline myopathy 10, 616165
LONP1	141.5	97.9	96.4	CODAS syndrome, 600373
LOXHD1	136.5	100	99.6	Deafness, autosomal recessive 77, 613079
LPAR6	104.2	99.7	98.2	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	134.6	99.8	97.8	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	111.5	100	99.6	Majeed syndrome, 609628
LPL	147.2	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LRAT	298.3	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	134.4	99.3	97.8	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	148.5	99.4	97.5	Urofacial syndrome 2, 615112
LRIT3	142.4	94.4	94.1	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	142.1	97.2	95.6	Albinism, oculocutaneous, type VII, 615179
LRP2	176.3	100	99.8	Donnai-Barrow syndrome, 222448
LRP4	166.6	99.1	98.9	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	189.8	98.2	97.9	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634

				Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRPAP1	138.3	99.5	97.2	Myopia 23, autosomal recessive, 615431
LRPPRC	127.3	99.4	97.2	Leigh syndrome, French-Canadian type, 220111
LRRC6	137.9	94.7	91	Ciliary dyskinesia, primary, 19, 614935
LRSAM1	130.4	100	99.7	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	125.9	99.8	96.6	Deafness, autosomal recessive 63, 611451
LSS	127.8	100	99.1	Cataract 44, 616509
LTBP2	104.6	99.6	97.1	?Weill-Marchesani syndrome 3, recessive, 614819 Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	113.5	98.7	94.7	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	117.1	98.6	95	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	54.3	71.8	61.4	Leukotriene C4 synthase deficiency, 614037
LYRM7	49	87.6	72.4	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	134.6	97.8	93.9	Chediak-Higashi syndrome, 214500
LZTFL1	109.1	99.1	95.3	Bardet-Biedl syndrome 17, 615994
MAB21L2	245.6	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAG	146.5	100	99.5	Spastic paraplegia 75, autosomal recessive, 616680
MAK	139.1	95.6	94.1	Retinitis pigmentosa 62, 614181
MALT1	136.6	89.1	85.4	Immunodeficiency 12, 615468
MAN1B1	128.9	100	99.7	Mental retardation, autosomal recessive 15, 614202
MAN2B1	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAPT	127.2	100	98.6	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540

				{Parkinson disease, susceptibility to}, 168600
MARS	125.2	99.7	97.3	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	173.2	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	159.4	97.5	94	Deafness, autosomal recessive 49, 610153
MASP1	148.6	100	99.6	3MC syndrome 1, 257920
MATN3	116.6	84.7	84.7	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600
MBOAT7	91.8	99.3	94.7	Mental retardation, autosomal recessive 57, 617188
MC2R	213.1	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCCC1	151.7	100	99.4	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	131.1	99.9	98.9	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	99.8	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	101.7	99.9	98.8	Factor V and factor VIII, combined deficiency of, 613625
MCM4	164.3	99.9	98.8	Immunodeficiency 54, 609981
MCM9	151.6	100	99.9	Ovarian dysgenesis 4, 616185
MCOLN1	150.2	98.8	97	Mucolipidosis IV, 252650
MCPH1	148.6	99.9	98.1	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	123.3	98	97.9	Epileptic encephalopathy, early infantile, 51, 617339
MECR	108.1	98.8	96.1	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED17	118	95.2	91.7	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	131.2	98.7	96.9	Mental retardation, autosomal recessive 18, 614249
MED25	103.9	99.1	95.7	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEFV	108.8	94.9	91	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	154.3	100	99.8	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	127.6	99.9	98.6	Carpenter syndrome 2, 614976
MEOX1	76.8	96.6	91.2	Klippel-Feil syndrome 2, 214300
MERTK	161.4	99.4	97.7	Retinitis pigmentosa 38, 613862
MESP2	81.5	93.1	87.9	Spondylocostal dysostosis 2, autosomal recessive, 608681

METTL23	145	100	100	Mental retardation, autosomal recessive 44, 615942
MFF	93.7	90.4	87.6	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	150.6	100	99.9	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	121.3	100	100	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	115.1	98.8	95.9	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	125.1	99.9	98.4	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	157.2	100	99.9	Congenital disorder of glycosylation, type IIa, 212066
MGME1	151.1	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	132	92.7	91.6	Keutel syndrome, 245150
MICU1	134.2	96	88.8	Myopathy with extrapyramidal signs, 615673
MKKS	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.5	99.9	98.5	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	103.4	100	99.8	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	162	100	99.7	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLYCD	75.6	91.3	86.9	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	183.2	100	99.6	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.2	100	99.9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	205.8	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	77	89.3	75	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	101.1	98.2	93.6	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP13	124.1	93.5	91.5	Metaphyseal anadysplasia 1, 602111

				Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP2	164.4	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100.5	100	98.6	Amelogenesis imperfecta, type IIA2, 612529
MMP21	93.3	90.2	84.6	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	125.4	96.2	91.8	Metaphyseal anadysplasia 2, 613073
MOCOS	169	99.1	96.6	Xanthinuria, type II, 603592
MOCS1	87.4	98.4	93	Molybdenum cofactor deficiency A, 252150
MOCS2	139.5	99.6	99.6	Molybdenum cofactor deficiency B, 252160
MOGS	121.6	99.8	99.1	Congenital disorder of glycosylation, type IIb, 606056
MPC1	121.8	100	99.5	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	111.8	100	99.7	Congenital disorder of glycosylation, type If, 609180
MPDZ	149	98.7	96.6	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	146.2	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPL	136.7	99.6	97.5	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	72.5	97.1	79.3	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	155	100	99.9	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	108.5	100	99.4	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRAP	161.3	100	100	Glucocorticoid deficiency 2, 607398
MRE11	51.2	95.3	82.3	Ataxia-telangiectasia-like disorder 1, 604391
MRPL3	66.3	91.2	77.9	Combined oxidative phosphorylation deficiency 9, 614582
MRPS16	161.1	100	99.1	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	166.4	99.7	97.9	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	138.8	95.3	91.8	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	132.7	99.9	98.3	Combined oxidative phosphorylation deficiency 32, 617664
MS4A1	123.7	99.4	96.2	Immunodeficiency, common variable, 5, 613495
MSH2	113.4	98.6	93.1	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320

MSH6	171.1	100	99.5	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSMO1	45.8	92.6	78.5	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	155.6	99.7	98.5	Deafness, autosomal recessive 74, 613718
MTFMT	124.6	99.3	96.2	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	126.1	98.4	97.2	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTMR2	106.6	100	99.2	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	173.7	89.5	87.3	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	109.6	98.9	93.5	?Spastic ataxia 4, autosomal recessive, 613672
MTR	140.9	99.8	98.8	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	139.1	100	99.2	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	132.4	99.9	98.8	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUSK	159.4	100	99.9	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	121.8	99.2	95.1	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	165	100	99.9	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVK	124.3	92.1	90.4	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYBPC1	150.7	99.9	99.4	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYD88	186.5	100	99.9	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260

MYH2	129.4	99.9	98.8	Proximal myopathy and ophthalmoplegia, 605637
MYL3	103.1	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYMK	141	100	100	Carey-Fineman-Ziter syndrome, 254940
MYO15A	116.5	97.3	94	Deafness, autosomal recessive 3, 600316
MYO18B	132.4	100	99.1	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	131.9	98.6	97	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	119.8	98.5	93	Deafness, autosomal recessive 30, 607101
MYO5A	125.3	99.5	97.4	Griscelli syndrome, type 1, 214450
MYO5B	137.6	98.2	95.5	Microvillus inclusion disease, 251850
MYO6	89.7	98.1	92.3	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	134.1	99.7	98.1	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYPN	142.4	99.3	98.4	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
NAGA	139.4	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	108.7	92.4	90.4	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	81	98	91.6	N-acetylglutamate synthase deficiency, 237310
NALCN	139.5	99.8	97.5	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	106.1	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	120	97.4	97.1	Combined oxidative phosphorylation deficiency 24, 616239
NBAS	145.3	99.5	97.6	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	172.7	99.5	99.3	Gray platelet syndrome, 139090
NBN	80.6	99.1	94.6	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065

				Nijmegen breakage syndrome, 251260
NCF1	23.9	25.8	22.1	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	124.3	100	99.4	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	158.7	100	100	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NDE1	100.9	100	99.5	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDRG1	128.4	99.9	98.8	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	201.4	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA11	86.9	99.5	95.8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160.2	100	100	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	133.9	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA9	124.7	98.6	93.2	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	115.6	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	58.6	85.7	70.9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	120.8	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	79.4	98.9	91.8	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	95.7	98.8	94.5	Mitochondrial complex I deficiency, nuclear type 16, 616238
NDUFAF6	79.3	97.1	85.6	Mitochondrial complex I deficiency, nuclear type 17, 612392
NDUFB11	109.6	94.4	88	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	22.6	91.9	59.2	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFS1	132.2	99.8	98.6	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	142.4	90.7	90.6	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	147.3	100	99.1	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	119.1	99.9	99.4	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	118.4	100	99.7	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	141.4	100	99.9	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	136.7	99.7	97.8	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	69.5	78.7	53.9	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	124	82.9	81.9	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	116.9	100	99.9	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	145.4	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060

NECTIN4	135.8	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK1	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK8	171.4	100	99.9	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	136.8	99.7	98.5	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	148.1	99.4	97.1	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROG3	119.4	100	99.3	Diarrhea 4, malabsorptive, congenital, 610370
NFU1	47.7	94.9	77.2	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	257.6	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	128	100	99.5	Congenital disorder of deglycosylation, 615273
NHEJ1	80.3	100	99.1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	174.2	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	143.5	99.7	98.6	?Seckel syndrome 7, 614851
NIPAL4	157.8	99.4	93.2	Ichthyosis, congenital, autosomal recessive 6, 612281
NKX2-6	104.4	100	99.7	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	55.8	92.4	73.9	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP2	119	100	99.8	No OMIM phenotype Beckwith-Wiedemann syndrome (Meyer (2009) PLoS Genet 5) Nijmegen breakage syndrome
NLRP7	135.9	99.8	98.7	Hydatidiform mole, recurrent, 1, 231090
NME8	105.6	97.5	91	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	137.9	100	99.7	Leber congenital amaurosis 9, 608553
NNT	136.9	98.6	97.1	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPC1	147.9	99.2	97.8	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	140.7	100	99.9	Niemann-pick disease, type C2, 607625

NPHP1	117.6	98.8	96.4	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	115.6	99.4	96.1	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	136.7	99.9	99.3	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	102	99.9	98.9	Nephrotic syndrome, type 1, 256300
NPHS2	116.9	99.8	95.9	Nephrotic syndrome, type 2, 600995
NPPA	115.4	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	164.8	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NR0B2	96.8	100	99.8	Obesity, mild, early-onset, 601665
NR1H4	141.9	96.6	92.2	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	93.4	99.9	98.9	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NRXN1	160.9	96.8	95.7	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSUN2	114.7	95.3	92.2	Mental retardation, autosomal recessive 5, 611091
NT5C2	125.3	97.1	92.7	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	62	89.3	78.8	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	166.3	100	99.8	Calcification of joints and arteries, 211800
NTHL1	98.1	99.1	95.7	Familial adenomatous polyposis 3, 616415
NTRK1	130.6	99.7	97.7	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUBPL	89.8	92.9	85.9	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP107	122.9	99.2	94.1	?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP62	111.6	100	99.9	Striatonigral degeneration, infantile, 271930
NUP93	140.7	97.9	94.9	Nephrotic syndrome, type 12, 616892
OAT	89.2	77.7	70.5	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870

OBSL1	140.5	99.8	98.5	3-M syndrome 2, 612921
OCA2	139.9	99.5	97.9	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OCLN	220.7	100	100	Pseudo-TORCH syndrome 1, 251290
OGDH	201.3	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA3	128	99.5	97.4	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	109.4	99.4	97.9	5-oxoprolinase deficiency, 260005
ORAI1	237.3	93.8	89.8	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	106.7	99.9	98.9	Meier-Gorlin syndrome 1, 224690
ORC4	57.6	95.8	82	Meier-Gorlin syndrome 2, 613800
ORC6	126.8	100	100	Meier-Gorlin syndrome 3, 613803
OSGEP	120.5	100	99.6	Galloway-Mowat syndrome 3, 617729
OSTM1	80.7	90.8	88.4	Osteopetrosis, autosomal recessive 5, 259720
OTOA	117.3	99	96.6	Deafness, autosomal recessive 22, 607039
OTOF	131.2	100	99.7	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	145.1	99.4	98.2	Deafness, autosomal recessive 18B, 614945
OTOGL	113.3	98.4	93.9	Deafness, autosomal recessive 84B, 614944
OTUD6B	123.4	99.9	98.2	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	149.5	90.5	86.3	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OXCT1	121.4	99.6	97.8	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RY12	186.2	100	100	Bleeding disorder, platelet-type, 8, 609821
P3H1	137	100	99.9	Osteogenesis imperfecta, type VIII, 610915
P3H2	100.2	99.2	93.4	Myopia, high, with cataract and vitreoretinal degeneration, 614292
PAH	151.7	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAM16	50.7	65.2	64.7	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	146.6	99.3	93.1	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	108.5	99.7	98.5	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847

PARK7	86.9	100	99	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PATL2	107.8	99.9	99.2	Oocyte maturation defect 4, 617743
PAX1	132.4	87.7	82.4	?Otofaciocervical syndrome 2, 615560
PC	149.3	97.7	94.6	Pyruvate carboxylase deficiency, 266150
PCBD1	113.3	99.5	99.1	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	103.1	96.4	89.2	Propionicacidemia, 606054
PCCB	129.8	98.7	96.4	Propionicacidemia, 606054
PCDH15	153.9	99	98	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCK1	143	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	194.5	100	99.8	PEPCK deficiency, mitochondrial, 261650
PCLO	165.2	99.7	98.5	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	117.6	98.9	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	147.2	100	99	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCYT1A	113.5	98.3	94.7	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE10A	119.8	81.2	80.8	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE6A	125.8	100	99.8	Retinitis pigmentosa 43, 613810
PDE6B	147.9	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	137	99	96.7	Cone dystrophy 4, 613093
PDE6G	95.3	99.5	96.3	Retinitis pigmentosa 57, 613582
PDE6H	68.4	97.6	77	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDHB	133	99.3	96.8	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.5	98.9	94.6	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	116.7	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	35.4	89	72.1	MODY, type IV, 606392

				Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDZD7	80.9	98.4	93.9	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	116	99.6	98.5	Prolidase deficiency, 170100
PET100	94.5	88.8	74.8	Mitochondrial complex IV deficiency, 220110
PEX1	115.8	97.7	95.4	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	111.8	96.1	90.1	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	105.7	99.7	98.3	?Peroxisome biogenesis disorder 14B, 614920
PEX12	168.3	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.6	99.8	98.7	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	149	99.7	97.5	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	137	97.1	93.1	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	92.9	99.9	99.2	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147.1	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	76.4	100	99.8	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	98.1	99.1	94.3	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	111.7	99.9	98.3	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	94.5	90.4	86.1	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	113.5	89.6	82	Peroxisome biogenesis disorder 9B, 614879

				Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	150.4	100	99.8	Glycogen storage disease VII, 232800
PGAM2	170.9	100	99.9	Glycogen storage disease X, 261670
PGAP1	98.1	94.9	88.6	Mental retardation, autosomal recessive 42, 615802
PGAP2	158.4	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	72.8	62.5	58	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGM1	133.6	100	99.9	Congenital disorder of glycosylation, type It, 614921
PGM3	191.4	99.9	99.7	Immunodeficiency 23, 615816
PHGDH	115.6	100	99.8	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKB	130.4	99.8	97.5	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	159.1	100	100	Cirrhosis due to liver phosphorylase kinase deficiency, 0 Glycogen storage disease IXc, 613027
PHOX2A	29.9	59.9	32.6	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	74.6	97.5	90.8	Refsum disease, 266500
PI4KA	112.8	93.6	89.4	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIEZO2	126.1	99.9	99.2	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PIGC	129	99.7	96.4	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	167.4	100	99.7	Mental retardation, autosomal recessive 53, 616917
PIGL	121.8	99.9	99.3	CHIME syndrome, 280000
PIGM	165.4	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	111.3	92.6	87.1	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147	100	99.9	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	171.3	98.1	98	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	145.5	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	147.6	100	99.8	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	121.4	100	99.9	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3R1	129.3	99.7	97.3	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880

PIK3R5	110.1	100	99.8	Ataxia-oculomotor apraxia 3, 615217
PINK1	90.3	87.2	81.1	Parkinson disease 6, early onset, 605909
PIP5K1C	107.6	96.3	95.1	Lethal congenital contractural syndrome 3, 611369
PKHD1	154.9	100	99.7	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	178.8	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	122	99.9	98.5	Ectodermal dysplasia/skin fragility syndrome, 604536
PLA2G6	117.5	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	125.6	99.9	97.2	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLCB1	142.8	100	99.7	Epileptic encephalopathy, early infantile, 12, 613722
PLCD1	116.9	99.5	97.1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	155.3	99.5	98.9	Nephrotic syndrome, type 3, 610725
PLEC	114.1	99.7	98.7	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	131.7	99.8	97.9	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	86.9	96.2	89	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	141.1	100	99.9	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	115.4	87.8	87	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLK4	145.5	99.5	96.3	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	137.9	99.8	97.5	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	108.6	94.7	88.6	Bruck syndrome 2, 609220
PLOD3	100.1	99	96.3	Lysyl hydroxylase 3 deficiency, 612394
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	120.8	99.4	96.8	Spinocerebellar ataxia, autosomal recessive 2, 213200

PMPCB	121	99.7	97.8	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	95.1	83.5	80.7	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PNKP	93	99.8	97.7	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	160.5	99.5	95.6	?Pancreatic lipase deficiency, 614338
PNP	151.4	100	99.5	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	192.6	100	100	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
PNPLA6	122.1	99.7	98.5	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPO	66.4	100	98.3	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	53.7	93.3	80.9	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	133.8	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	78.7	98	94.2	Cone-rod dystrophy 20, 615973
POLG	114.4	100	99.5	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLH	140.7	100	99.8	Xeroderma pigmentosum, variant type, 278750
POLR1C	117	99.7	96.1	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR3A	137.4	100	99.9	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	146.4	99.9	98.5	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	116.2	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	127.6	99.7	97.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151

				Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123
POMGNT2	259.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	205.1	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	114.4	95.2	87.5	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POMT1	155.7	99.7	98.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	111.1	98.9	97.5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POR	167.7	99.9	98.7	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	106.3	98.2	94.7	Pituitary hormone deficiency, combined, 1, 613038
PPIB	118.4	100	100	Osteogenesis imperfecta, type IX, 259440
PPP1R15B	133.4	99.4	98	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPT1	144.5	90	87.3	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	89.4	100	99.9	Retinitis pigmentosa 36, 610599
PRDM12	112.2	91	87.7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM5	129.4	99.5	95.5	Brittle cornea syndrome 2, 614170
PRF1	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	144.6	97.6	88.4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRKCD	181.2	100	99.9	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	106.7	98.4	94.8	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKN	98.6	79.6	78.8	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572

PRKRA	179.6	99.8	98.4	Dystonia 16, 612067
PRMT7	138.7	100	99.8	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROC	138.5	99.7	97.2	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	83.8	84.9	82.3	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROM1	112.3	95.4	92.8	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	76.5	91.6	84.3	Pituitary hormone deficiency, combined, 2, 262600
PROS1	101.4	96.8	91.4	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRSS12	153.3	99.9	98.5	Mental retardation, autosomal recessive 1, 249500
PRSS56	51.5	96.6	83.8	Microphthalmia, isolated 6, 613517
PRX	115.6	99.8	98.3	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	114.4	99.9	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.2	91.4	75.8	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSMB8	118.7	100	99.8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMC3IP	113.4	99.9	99.7	Ovarian dysgenesis 3, 614324
PSPH	128.9	98.8	95.4	Phosphoserine phosphatase deficiency, 614023
PTF1A	74	88.4	78.1	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTH1R	108.5	99.9	98.8	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPN14	175.7	99.4	96.4	?Choanal atresia and lymphedema, 613611
PTPRC	101.6	93.9	86.3	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971

				{Hepatitis C virus, susceptibility to}, 609532
PTPRO	140.8	99.9	99	Nephrotic syndrome, type 6, 614196
PTPRQ	104.7	93.3	89.1	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	279.6	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	107.2	99.6	94.1	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	127.2	98.6	93.9	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	192	100	100	?Mental retardation, autosomal recessive 55, 617051
PUS7	136.5	99.8	98	No OMIM phenotype ?Intellectual disability (Riazuddin (2017) Mol Psychiatry 22,1604)
PXDN	163.8	99.8	98.5	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	86.3	99.4	94.3	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	127.6	100	97.6	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	158.6	100	100	Glycogen storage disease VI, 232700
PYGM	127.1	100	99.9	McArdle disease, 232600
QARS	166.6	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	92.3	100	99.5	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	82.7	97.1	86.4	Warburg micro syndrome 3, 614222
RAB23	110.3	99.7	98	Carpenter syndrome, 201000
RAB27A	143.9	100	99.9	Griscelli syndrome, type 2, 607624
RAB28	52.1	96.5	87.1	Cone-rod dystrophy 18, 615374
RAB33B	233.9	100	100	Smith-McCort dysplasia 2, 615222
RAB3GAP1	124.2	99.4	98.8	Warburg micro syndrome 1, 600118
RAB3GAP2	94.1	98.4	93.9	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAD50	99	92.6	86.2	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	143.4	100	98.9	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAG1	206.9	100	100	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457

RAG2	221	100	100	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAPSN	140.5	99.6	96.3	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	118.7	100	100	Microphthalmia, syndromic 12, 615524
RARS	86.4	92.7	85.9	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	107.2	100	99.1	Pontocerebellar hypoplasia, type 6, 611523
RAX	82.5	88.7	77.3	Microphthalmia, isolated 3, 611038
RBBP8	110.7	99.6	96.4	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744
RBCK1	104.1	99.2	94.9	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM28	138.7	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	106.6	100	99.4	Thrombocytopenia-absent radius syndrome, 274000
RBP4	99.6	99.4	96.1	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	123.7	100	99.7	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	161.6	100	99.9	Leber congenital amaurosis 12, 610612
RDH12	94.4	100	98.4	Leber congenital amaurosis 13, 612712
RDH5	160.2	100	99.7	Fundus albipunctatus, 136880
RDX	43.2	84.7	64.8	Deafness, autosomal recessive 24, 611022
RECQL4	149.6	99.2	96.5	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RELN	155.6	100	99.8	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	148.9	100	100	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
RETREG1	126.1	95.6	90.1	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFT1	108.3	99.8	97.3	Congenital disorder of glycosylation, type In, 612015
RFX5	116.9	98.7	96.3	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920

RFX6	153.9	100	99.6	Mitchell-Riley syndrome, 615710
RFXANK	105.9	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	84.8	94.4	91.6	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	126.8	100	99.4	Retinitis pigmentosa 44, 613769
RHO	210.1	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIN2	113.4	100	99.9	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	163.3	100	99.6	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RLBP1	144.8	100	100	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RMND1	137.2	99.8	97.3	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	98.6	99.1	95.6	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	142.1	100	99.9	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93.2	87.5	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.2	100	99.9	Aicardi-Goutieres syndrome 3, 610329
RNASET2	96.4	91.9	88.3	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF168	215.3	100	99.1	RIDDLE syndrome, 611943
RNF216	137.1	99.8	98.6	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
ROBO3	99.7	98.7	95.6	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	112.2	97.9	95.3	Kohlschutter-Tonz syndrome, 226750
ROR2	165.9	99.4	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	132.3	100	100	Immunodeficiency 42, 616622
RP1	121.8	91.4	90.3	Retinitis pigmentosa 1, 180100
RPE65	130.3	100	99.3	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGRIP1	154.2	100	99.9	Cone-rod dystrophy 13, 608194

				Leber congenital amaurosis 6, 613826
RPGRIP1L	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPIA	113.1	94.3	90.9	?Ribose 5-phosphate isomerase deficiency, 608611
RRM2B	128.6	99.7	97.5	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RSPH1	146.1	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	132.5	99.7	97.5	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	139	98.3	96.5	Ciliary dyskinesia, primary, 11, 612649
RSPH9	127.4	100	99.6	Ciliary dyskinesia, primary, 12, 612650
RSPO1	109.7	100	100	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	107.2	100	100	Anonychia congenita, 206800
RSPRY1	168.9	100	99.9	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RTEL1	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN4IP1	98.1	99.9	99.1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	129.5	97.2	94.7	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	104.1	98	97.5	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	182.3	100	99.9	Mental retardation, autosomal recessive 61, 617773
RYR1	120.7	96.8	93.7	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
S1PR2	278	97.4	92.7	Deafness, autosomal recessive 68, 610419
SACS	154.5	100	99.7	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	131.3	100	99.9	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SAMD9	159.1	99.9	99.3	MIRAGE syndrome, 617053

				Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	127.9	99.6	96.6	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	125	89.8	88.9	Chylomicron retention disease, 246700
SARS2	104.8	94.8	92.7	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SBDS	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	107.9	98.5	96.5	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	117	99.6	96.8	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	198.4	100	99.2	Lathosterolosis, 607330
SCAPER	135.9	96	93.6	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	121.2	100	99.9	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	82.5	85.2	73.6	Van den Ende-Gupta syndrome, 600920
SCN1B	168.3	97.1	96.1	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN4A	214	99.9	99.5	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SCN9A	146.5	98.5	97	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder,, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	134.3	99	96.3	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	148.9	100	99.8	Bronchiectasis with or without elevated sweat chloride 1, 211400

				Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	139.4	99.7	97.1	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	109.6	97.9	94.3	Mitochondrial complex IV deficiency, 220110
SCO2	113.1	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	106.9	99.6	96.5	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	143.2	98.6	96.3	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	123.9	99.8	97.4	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	42.5	100	96.2	Mitochondrial complex II deficiency, 252011
SEC23A	121.8	98	94.6	Craniolenticulosutural dysplasia, 607812
SEC23B	161.1	97.5	96.4	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	136.9	99.9	98.6	Cole-Carpenter syndrome 2, 616294
SECISBP2	122.5	98.3	95.3	Thyroid hormone metabolism, abnormal, 609698
SELENON	111.7	85.2	83.3	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEPSECS	159.3	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	141.1	100	99.9	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
SERPINA6	190.9	100	100	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	164.1	95.9	95.9	?Deafness, autosomal recessive 91, 613453
SERPINB7	127.4	100	99.5	Palmoplantar keratoderma, Nagashima type, 615598
SERPINC1	143.1	100	100	Thrombophilia due to antithrombin III deficiency, 613118

SERPINE1	155	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	101.8	100	99.4	Osteogenesis imperfecta, type VI, 613982
SERPINF2	143.8	99.9	99.3	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	97.9	97.3	92.6	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SETX	163.2	99.9	99.1	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SFRP4	164.1	100	98.5	Pyle disease, 265900
SFTPB	99.4	99.9	98.7	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFXN4	131.7	100	99.1	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	147.3	100	99.7	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	154.2	96.6	94.2	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94.8	100	99.4	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	138.7	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	94.8	98	94.3	Chronic atrial and intestinal dysrhythmia, 616201
SGSH	129	95.1	93.6	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	140.2	100	99.8	Frank-ter Haar syndrome, 249420
SH3TC2	121.3	100	99.7	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SI	98.1	96.2	87.1	Sucrase-isomaltase deficiency, congenital, 222900
SIL1	154.4	99.8	98	Marinesco-Sjogren syndrome, 248800
SIX6	228.6	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIV2L	149.1	100	99.8	Trichohepatoenteric syndrome 2, 614602
SLC10A2	151	100	100	Bile acid malabsorption, primary, 613291
SLC11A2	135.2	100	99.3	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	172.7	99.8	99.1	Bartter syndrome, type 1, 601678
SLC12A3	139.3	100	100	Gitelman syndrome, 263800
SLC12A5	125.9	85.4	81.6	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	141.8	100	99.9	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	164.1	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC17A5	119.6	96.8	92.9	Salla disease, 604369

				Sialic acid storage disorder, infantile, 269920
SLC19A2	119.5	99.8	97.8	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	186.4	100	99.9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	174.2	100	99.9	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A4	156.5	98.9	94.9	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	105.7	100	99.7	Hypouricemia, renal, 220150
SLC22A5	153.3	100	100	Carnitine deficiency, systemic primary, 212140
SLC24A1	218.5	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	126.8	99.6	97.3	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	114.5	99.6	97.7	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	71	92.2	87	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	150.5	99.8	98.4	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	110.7	95.7	92.3	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	192.5	98.8	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	88.6	99.9	98.3	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	110.3	100	99.7	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	108.7	99.5	96.9	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A26	98	98.9	96	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	139	99.8	97.6	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	111.4	99.8	98.1	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A46	205.7	95.9	87.3	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	139.8	100	99.9	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	233.2	100	100	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600

				Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	156.1	99.9	98.9	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	123.3	99.9	99.1	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	150.9	98.7	95.8	?Deafness, autosomal recessive 61, 613865
SLC27A4	155.8	99.6	97.9	Ichthyosis prematurity syndrome, 608649
SLC29A3	203.6	99.9	99.5	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	190.1	92.9	92.8	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	166.4	97.7	97.6	Arterial tortuosity syndrome, 208050
SLC2A2	178.4	100	99.9	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	119	99.2	96.2	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	164.4	100	100	Hypermanganesemia with dystonia 1, 613280
SLC33A1	140.9	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	153.2	100	99.5	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	169.1	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	105.6	98.9	94.5	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	124	99.9	97.7	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	230.2	99.9	98.4	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	115.4	95.7	90.4	Schneckenbecken dysplasia, 269250
SLC37A4	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	76.4	99.4	95.8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	114.8	99.8	98	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350

SLC39A14	107.7	99.8	97.9	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	81.8	99.2	96	Acrodermatitis enteropathica, 201100
SLC39A8	128.5	100	99.7	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	162.8	100	99.5	Cystinuria, 220100
SLC45A2	148.1	100	99.9	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	106	99.4	96.4	Folate malabsorption, hereditary, 229050
SLC4A1	140	100	99.9	Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653 [Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Malaria, resistance to], 611162
SLC4A11	153	100	99.7	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700
SLC4A4	122.3	99	97.1	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A2	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.6	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	140	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	118.7	100	100	Renal glucosuria, 233100
SLC5A5	93.9	99.7	96.8	Thyroid dyshormonogenesis 1, 274400
SLC6A17	189.8	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	149.1	100	99.3	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600

SLC6A3	145.7	100	99.8	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	136.5	100	99.7	Hyperekplexia 3, 614618
SLC7A14	191.9	100	100	Retinitis pigmentosa 68, 615725
SLC7A7	123.9	100	99.9	Lysinuric protein intolerance, 222700
SLC7A9	125.5	99.9	99	Cystinuria, 220100
SLC9A3	147.1	98.7	96.9	Diarrhea 8, secretory sodium, congenital, 616868
SLCO2A1	110.5	100	99.6	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK6	206.9	100	100	Deafness and myopia, 221200
SLURP1	97.1	99.8	96.2	Meleda disease, 248300
SLX4	114.2	100	99.8	Fanconi anemia, complementation group P, 613951
SMARCAL1	134.6	100	99.9	Schimke immunoosseous dysplasia, 242900
SMG9	101.8	100	99.9	Heart and brain malformation syndrome, 616920
SMN1	112.7	99.8	96.5	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMOC1	129.8	99.5	97.3	Microphthalmia with limb anomalies, 206920
SMOC2	91.5	75.4	72.6	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	123.5	99.6	97.9	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNAI2	129.8	100	99.8	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	153.5	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	139.3	99.2	96.7	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNX10	118.9	96.2	96.1	Osteopetrosis, autosomal recessive 8, 615085
SNX14	70.1	95.2	82.9	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	130.7	92.9	85.4	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	161.9	100	100	Amyotrophic lateral sclerosis 1, 105400
SOST	112.6	100	99.6	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX18	21.2	62.6	48.8	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940

SP110	121.6	100	99.5	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPAG1	87.3	96.3	88.2	Ciliary dyskinesia, primary, 28, 615505
SPARC	161	100	100	Osteogenesis imperfecta, type XVII, 616507
SPART	132.4	99.8	98.2	Troyer syndrome, 275900
SPATA5	132	99.9	99.2	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	119.6	97.8	90.8	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPEG	100.9	93.3	86.5	Centronuclear myopathy 5, 615959
SPG11	129.2	99.2	96.9	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	121.4	98.6	94.8	Mast syndrome, 248900
SPG7	119.2	93.3	92.4	Spastic paraplegia 7, autosomal recessive, 607259
SPINK5	145	99.4	96.5	Netherton syndrome, 256500
SPINT2	71.5	97.8	84.2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	166.5	98.9	90	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRTN	167.8	100	100	Ruijs-Aalfs syndrome, 616200
SPTA1	119.4	99.8	98.9	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTB	148	100	99.9	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649
SPTBN2	118	99.9	99.3	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SRD5A2	77.6	100	96.4	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	135.9	100	99.7	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST14	154	98	97	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	144.5	100	99.9	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	121.9	84.4	84.2	Salt and pepper developmental regression syndrome, 609056
STAC3	120	100	99.9	Myopathy, congenital, Baily-Bloch, 255995

STAMBP	112.3	99.3	96.5	Microcephaly-capillary malformation syndrome, 614261
STAR	124	100	100	Lipoid adrenal hyperplasia, 201710
STAT1	126.2	98	95.8	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	116	100	99.9	Immunodeficiency 44, 616636
STAT5B	130.6	99.7	97.2	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIL	157.2	99.8	98.6	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK4	138.9	100	99.3	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STRA6	116.5	100	99.9	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	134.1	100	98.9	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	119	99.9	98.4	Deafness, autosomal recessive 16, 603720
STT3A	156.2	100	100	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	125.1	99.2	96	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	176.1	100	98.9	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	311.4	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	102.3	88.9	83.8	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	64.9	93.3	82.8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	101.3	99.6	95.4	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	122.6	99.9	99	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	103.3	98.6	91.1	Multiple sulfatase deficiency, 272200
SUOX	212.6	100	100	Sulfite oxidase deficiency, 272300
SURF1	96.2	88.3	88.3	Charcot-Marie-Tooth disease, type 4K, 616684

				Leigh syndrome, due to COX IV deficiency, 256000
SYNE1	136.6	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE4	73.9	98.3	91.6	Deafness, autosomal recessive 76, 615540
SYNJ1	127.2	99.3	96.1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYT14	113.5	59.9	53.8	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	149.5	99.5	99.2	Epileptic encephalopathy, early infantile, 18, 615476
TAC3	80.5	99.1	91.1	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	91.7	97	92.6	Mitochondrial complex IV deficiency, 220110
TACR3	180.3	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	223.6	99.1	96.6	Corneal dystrophy, gelatinous drop-like, 204870
TAF13	81.8	100	99.9	Mental retardation, autosomal recessive 60, 617432
TAF2	112.8	98.8	94.7	Mental retardation, autosomal recessive 40, 615599
TAF6	130	99.9	98.6	Alazami-Yuan syndrome, 617126
TALDO1	130.5	100	99.9	Transaldolase deficiency, 606003
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	103.3	100	99.1	Bare lymphocyte syndrome, type I, 604571
TAP2	95.2	99.6	98.6	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	100.7	96.1	94.3	Bare lymphocyte syndrome, type I, 604571
TAPT1	83.5	88.5	85.9	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TAT	143.1	100	100	Tyrosinemia, type II, 276600
TBC1D20	145.7	94.2	94.1	Warburg micro syndrome 4, 615663
TBC1D23	86	95.7	91.5	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	179.2	100	100	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	105.5	99.6	96.6	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	152.9	95.5	92.3	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	128	99.9	98.2	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410

				Kenny-Caffey syndrome, type 1, 244460
TBCK	86.5	95.7	89.3	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBX15	131.7	100	99.4	Cousin syndrome, 260660
TBX19	174.4	100	100	Adrenocorticotropic hormone deficiency, 201400
TBXAS1	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCAP	89	100	99.2	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCIRG1	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TCN2	175.6	100	100	Transcobalamin II deficiency, 275350
TCTN1	98.8	95.7	92.8	Joubert syndrome 13, 614173
TCTN2	144.2	99.5	97	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDP1	122.9	98.7	95.3	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	165.1	99.9	98.8	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	168.3	99	97.7	Cataract 36, 613887
TECPR2	161.1	100	99.9	Spastic paraplegia 49, autosomal recessive, 615031
TECR	94.6	99.9	97.9	Mental retardation, autosomal recessive 14, 614020
TECTA	208	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TELO2	98.1	97.4	93.7	You-Hoover-Fong syndrome, 616954
TENM3	185.7	99.5	98.7	Microphthalmia, isolated, with coloboma 9, 615145
TF	125.9	100	100	Atransferrinemia, 209300
TFR2	104.4	98.2	93.5	Hemochromatosis, type 3, 604250
TFRC	157.2	99.9	99.1	Immunodeficiency 46, 616740
TG	133.4	100	99.7	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	82.4	98.1	88.8	Catel-Manzke syndrome, 616145
TGM1	158.8	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	173.9	100	100	Peeling skin syndrome 2, 609796
TH	68.2	97.6	88.7	Segawa syndrome, recessive, 605407
THOC6	248.6	100	99.9	Beaulieu-Boycott-Innes syndrome, 613680

THRB	167.4	100	99.5	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TJP2	111.1	93.8	92.2	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	105.7	93.4	89.4	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	114.1	98.7	97.7	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	109.9	99.9	98.9	Preimplantation embryonic lethality, 616814
TMC1	122.8	98.2	93.8	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	83.7	99.9	99	Epidermodysplasia verruciformis, 226400
TMC8	108.1	97.6	91.6	Epidermodysplasia verruciformis 2, 618231
TMCO1	78.7	88	86.5	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM126A	120.3	98.4	86.2	Optic atrophy 7, 612989
TMEM126B	79.2	99.8	97.7	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM138	100.2	100	99.5	Joubert syndrome 16, 614465
TMEM165	113.9	99.8	98.1	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	105.1	100	99.9	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	111.9	100	98.7	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.7	99.8	98.3	Joubert syndrome 14, 614424
TMEM38B	114.3	100	99.4	Osteogenesis imperfecta, type XIV, 615066
TMEM5	120.5	96.8	92.9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	72.9	93.3	83.4	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	138.7	94.6	90.3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052

TMIE	109.6	98.8	92.1	Deafness, autosomal recessive 6, 600971
TMPRSS15	116.9	95.5	89.1	Enterokinase deficiency, 226200
TMPRSS3	125.5	100	99.9	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	101.7	100	99.1	Iron-refractory iron deficiency anemia, 206200
TNFRSF11B	224.8	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	102.1	100	99.7	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	55.8	76.5	66.8	Immunodeficiency, common variable, 4, 613494
TNFSF11	150.4	99.3	93.2	Osteopetrosis, autosomal recessive 2, 259710
TNIK	111.1	99.9	99.3	Mental retardation, autosomal recessive 54, 617028
TNNI3	86.7	98.1	86.5	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNT1	86.9	96.3	94	Nemaline myopathy 5, Amish type, 605355
TNXB	96.4	98.4	91.4	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	165.1	100	100	Pontocerebellar hypoplasia, type 7, 614969
TP53RK	37.4	91.3	76.5	Galloway-Mowat syndrome 4, 617730
TPI1	103	99.2	96.7	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	112.7	99.8	97.3	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPO	134.8	99.9	98.5	Thyroid dyshormonogenesis 2A, 274500
TPP1	146.3	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRN	62.7	74.7	65.4	Deafness, autosomal recessive 79, 613307
TRAC	170.9	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	90.3	96.3	92.8	Senior-Loken syndrome 9, 616629
TRAIP	141.6	100	100	Seckel syndrome 9, 616777
TRAPPC11	126.2	99.4	96.4	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC9	135	100	99.9	Mental retardation, autosomal recessive 13, 613192
TRDN	71.9	83.6	70.8	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	149	99.9	99.6	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	242.4	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448

				Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRH	75.8	95.9	85.1	Thyrotropin-releasing hormone deficiency, 275120
TRIM2	157.7	93.6	91.4	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	110.2	98.2	97.2	Mulibrey nanism, 253250
TRIOBP	135.6	97	94.9	Deafness, autosomal recessive 28, 609823
TRIP11	84.3	95.2	87.4	Achondrogenesis, type IA, 200600
TRIP4	113.5	100	98.8	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRMT10A	135.2	100	99.4	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	131.4	99.8	98.8	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	208.7	99.2	93.9	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99	100	99.6	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	104.6	97.8	92.3	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPM1	161	100	99.6	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	151.1	99.8	98.7	Hypomagnesemia 1, intestinal, 602014
TSEN15	74.2	99	93.6	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	123.8	100	99.8	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	53.5	90.5	85.7	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	82.9	95.9	92.9	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	127.2	100	100	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	271.7	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	216.5	99.2	96.8	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSPAN12	129.4	100	99.5	Exudative vitreoretinopathy 5, 613310

TSPEAR	141.5	100	99	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	141.5	100	99.4	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTC19	92.1	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	100.7	99.7	97.6	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	103.4	100	99.5	Ciliary dyskinesia, primary, 35, 617092
TTC37	124	99.6	98.1	Trichohepatoenteric syndrome 1, 222470
TTC7A	123	99.9	98.3	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	106.9	97.9	92	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	104.5	100	99.7	Mental retardation, autosomal recessive 39, 615541
TTLL5	152.7	99.9	98.7	Cone-rod dystrophy 19, 615860
TTPA	101.5	83.6	76.6	Ataxia with isolated vitamin E deficiency, 277460
TUBA8	177.1	99.9	99.7	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBGCP4	130.8	99.1	96.2	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	152.2	99.9	98.9	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	135.4	100	99.7	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	97.8	96.8	91.7	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	136.4	100	98.3	Mental retardation, autosomal recessive 7, 611093
TWIST2	131.3	100	99.3	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	178.8	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
TXNL4A	109	100	99.5	Burn-McKeown syndrome, 608572
TYK2	119.2	99.9	98.8	Immunodeficiency 35, 611521
TYMP	95.2	98.3	85	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	185.3	100	100	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470

				[Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYROBP	95.2	100	99.9	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	176.9	100	99.9	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA5	75.4	94.1	77.1	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2T	107	100	99.3	Fanconi anemia, complementation group T, 616435
UBE3B	127.8	100	99.9	Kaufman oculocerebrofacial syndrome, 244450
UBR1	128.2	99.2	96	Johanson-Blizzard syndrome, 243800
UGT1A1	240.6	100	100	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMPS	173.6	99.3	97.2	Orotic aciduria, 258900
UNC13D	97	99.6	97.7	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	133.4	99.9	99.1	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	78.4	99.5	94.2	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	157.4	100	100	Beta-ureidopropionase deficiency, 613161
UQCRB	107.6	99.6	96.8	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	122.6	99.9	99.1	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	131.3	100	99.9	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	132	99.9	99	?Urocanase deficiency, 276880
UROD	163.1	99.8	97.9	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	108.3	100	99.9	Porphyria, congenital erythropoietic, 263700
USB1	125	99.9	98.2	Poikiloderma with neutropenia, 604173
USH1C	97.5	100	99.4	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	195.3	98.4	96.3	Usher syndrome, type 1G, 606943
USH2A	148.5	100	99.7	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901

UVSSA	149.4	99.1	98.4	UV-sensitive syndrome 3, 614640
VAC14	108.4	99.8	98.5	Striatonigral degeneration, childhood-onset, 617054
VARS2	110.9	99.9	98.9	Combined oxidative phosphorylation deficiency 20, 615917
VDR	123.3	98	95.2	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VHL	119.7	92.6	85.3	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIPAS39	144.6	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	162.1	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	200.9	99.9	99.4	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	144.9	95.3	93.2	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	69.5	95.3	85.3	Choreoacanthocytosis, 200150
VPS13B	143.8	98.6	96.8	Cohen syndrome, 216550
VPS13C	106.6	96.6	90	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS33B	138.3	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	73.6	86.6	66.4	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	131.5	96.2	94.9	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	129.2	91.4	90.4	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	124.8	97.5	94.2	Pontocerebellar hypoplasia type 1A, 607596
VSX2	77.7	99.8	97.3	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	141.5	99.8	98.4	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WASHC4	91.8	95.3	89.6	?Mental retardation, autosomal recessive 43, 615817
WDPCP	107.3	93.9	88.9	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR34	106.6	99.5	96.2	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	161.5	100	99.7	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317

WDR72	132.2	96.5	95.4	Amelogenesis imperfecta, type IIA3, 613211
WDR73	138.9	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	163.3	99.9	99.4	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	102.2	99.6	95.7	Oocyte maturation defect 5, 617996
WFS1	251.4	100	99.7	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	114	99.8	98.8	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WISP3	118.4	100	100	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	167.7	99.9	99.5	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNT1	188.8	100	99.9	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	114	100	99.1	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	144.7	100	99.9	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	166.8	100	99.6	?Tetra-amelia syndrome 1, 273395
WNT4	263.1	93.4	92.7	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT7A	216.8	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	123.6	98.3	94.6	Werner syndrome, 277700
WWOX	130.9	100	99.7	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	109.1	100	99.9	Xanthinuria, type I, 278300

XPA	52.9	98.5	88.9	Xeroderma pigmentosum, group A, 278700
XPC	140.7	100	99.7	Xeroderma pigmentosum, group C, 278720
XPNPEP3	134	100	99.2	Nephronophthisis-like nephropathy 1, 613159
XRCC4	103.2	99.7	97.3	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	132.5	90.4	87.1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.3	98.9	94.9	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YARS2	173.2	99.8	98.9	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	105.3	97.7	91.9	?Optic atrophy 11, 617302
YY1AP1	159.2	98.3	97	Grange syndrome, 602531
ZAP70	185.6	99.9	99.5	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB16	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB24	178.1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	184.1	99.6	97.2	Mental retardation, autosomal recessive 56, 617125
ZFYVE26	120.3	99.9	99.4	Spastic paraplegia 15, autosomal recessive, 270700
ZMPSTE24	113.3	100	99.1	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	136.5	100	100	Ciliary dyskinesia, primary, 22, 615444
ZNF408	135.7	100	100	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	250.8	100	100	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	93.1	98.7	96.3	Brittle cornea syndrome 1, 229200
ZNF513	110.7	100	99.7	?Retinitis pigmentosa 58, 613617
ZP1	196	100	100	Oocyte maturation defect 1, 615774

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 31st. 2018.

This list is accurate for panel version DG 2.15

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