INTELLECTUAL DISABILITY GENE PANEL DG 3.4.0 (1612 genes)

Releasedate: 19-04-2022

Gene	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AAAS	100,0%	100,0%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100,0%	100,0%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287 ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 Trichothiodystrophy 8, nonphotosensitive, 619691
AASS	100,0%	100,0%	Hyperlysinemia, 238700
ABAT	100,0%	100,0%	GABA-transaminase deficiency, 613163
ABCA2	100,0%	100,0%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCC8	100,0%	100,0%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	100,0%	100,0%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 ?Atrial fibrillation, familial, 12, 614050 Intellectual disability and myopathy syndrome, 619719
ABCD1	100,0%	100,0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD16A	100,0%	100,0%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	100,0%	100,0%	Chanarin-Dorfman syndrome, 275630
ACAD9	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	100,0%	100,0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100,0%	100,0%	2-methylbutyrylglycinuria, 610006
ACAT1	100,0%	100,0%	Alpha-methylacetoacetic aciduria, 203750
ACER3	100,0%	100,0%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	100,0%	100,0%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559

ACOX1	100,0%	100,0%	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	100,0%	100,0%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100,0%	100,0%	Intellectual developmental disorder, X-linked 63, 300387
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	100,0%	100,0%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACTL6A	100,0%	100,0%	No OMIM Disease ID
ACTL6B	100,0%	100,0%	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACVR1	100,0%	100,0%	Fibrodysplasia ossificans progressiva, 135100
ACY1	100,0%	100,0%	Aminoacylase 1 deficiency, 609924
ADAM22	100,0%	100,0%	Developmental and epileptic encephalopathy 61, 617933
ADAR	100,0%	100,0%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADARB1	95,1%	95,1%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100,0%	100,0%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADD3	100,0%	100,0%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100,0%	100,0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADK	84,5%	84,5%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	95,4%	95,4%	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100,0%	100,0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	100,0%	100,0%	Adenylosuccinase deficiency, 103050
AFF2	100,0%	99,9%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	100,0%	100,0%	KINSSHIP syndrome, 619297
AFF4	100,0%	100,0%	CHOPS syndrome, 616368
AFG3L2	100,0%	100,0%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGA	100,0%	100,0%	Aspartylglucosaminuria, 208400
AGAP1	100,0%	100,0%	No OMIM Disease ID
AGMO	100,0%	100,0%	No OMIM Disease ID
AGO1	100,0%	100,0%	No OMIM Disease ID
AGO2	100,0%	99,9%	Lessel-Kreienkamp syndrome, 619149
AGTPBP1	100,0%	100,0%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276

AHCY	100,0%	100,0%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100,0%	100,0%	Xia-Gibbs syndrome, 615829
AHI1	100,0%	100,0%	Joubert syndrome 3, 608629
AHSG	100,0%	100,0%	?Alopecia-intellectual disability syndrome 1, 203650
AIFM1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
AIMP1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100,0%	100,0%	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	100,0%	100,0%	Spastic paraplegia 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603
ALDH3A2	93,2%	93,2%	Sjogren-Larsson syndrome, 270200
ALDH4A1	100,0%	100,0%	Hyperprolinemia, type II, 239510
ALDH5A1	100,0%	100,0%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	100,0%	100,0%	Epilepsy, pyridoxine-dependent, 266100
ALG1	100,0%	100,0%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,8%	96,8%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100,0%	100,0%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100,0%	99,9%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100,0%	100,0%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	100,0%	100,0%	Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100,0%	100,0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100,0%	100,0%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96,6%	96,6%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100,0%	100,0%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100,0%	100,0%	Alstrom syndrome, 203800
ALX3	100,0%	100,0%	Frontonasal dysplasia 1, 136760

ALX4	100,0%	100,0%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMER1	100,0%	100,0%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100,0%	100,0%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	100,0%	100,0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100,0%	100,0%	Glycine encephalopathy, 605899
ANK2	100,0%	100,0%	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 37, 615493
ANKH	100,0%	100,0%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	100,0%	100,0%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	100,0%	100,0%	KBG syndrome, 148050
ANKRD17	100,0%	100,0%	Chopra-Amiel-Gordon syndrome, 619504
ANKS1B	100,0%	100,0%	No OMIM Disease ID
ANO10	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	100,0%	100,0%	GAPO syndrome, 230740
AP1G1	100,0%	100,0%	Usmani-Riazuddin syndrome, autosomal recessive, 619548 Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100,0%	100,0%	MEDNIK syndrome, 609313
AP1S2	99,9%	99,9%	Pettigrew syndrome, 304340
AP2M1	100,0%	100,0%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	100,0%	100,0%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	100,0%	100,0%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100,0%	99,7%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100,0%	100,0%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100,0%	100,0%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100,0%	100,0%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100,0%	100,0%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	87,9%	87,9%	Spastic paraplegia 52, autosomal recessive, 614067
APC2	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 10, 618677 Intellectual developmental disorder, autosomal recessive 74, 617169
APTX	100,0%	100,0%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	97,0%	96,6%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	100,0%	100,0%	Periventricular nodular heterotopia 8, 618185

ARF3	100,0%	100,0%	No OMIM Disease ID
ARFGEF1	100,0%	100,0%	No OMIM Disease ID
ARFGEF2	100,0%	100,0%	Periventricular heterotopia with microcephaly, 608097
ARG1	92,9%	92,9%	Argininemia, 207800
ARHGAP31	100,0%	100,0%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100,0%	100,0%	No OMIM Disease ID
ARHGEF6	100,0%	100,0%	No OMIM Disease ID
ARHGEF9	97,2%	97,2%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100,0%	100,0%	Coffin-Siris syndrome 2, 614607
ARID1B	98,6%	98,3%	Coffin-Siris syndrome 1, 135900
ARID2	100,0%	100,0%	Coffin-Siris syndrome 6, 617808
ARL13B	100,0%	100,0%	Joubert syndrome 8, 612291
ARL6	100,0%	100,0%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC9	100,0%	100,0%	Joubert syndrome 30, 617622
ARPC4	100,0%	100,0%	No OMIM Disease ID
ARSA	100,0%	100,0%	Metachromatic leukodystrophy, 250100
ARSL	100,0%	100,0%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100,0%	100,0%	Developmental and epileptic encephalopathy 38, 617020
ARX	99,0%	96,8%	Proud syndrome, 300004 Hydranencephaly with abnormal genitalia, 300215 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Lissencephaly, X-linked 2, 300215 Intellectual developmental disorder, X-linked 29, 300419
ASAH1	100,0%	100,0%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASH1L	98,7%	98,7%	Intellectual developmental disorder, autosomal dominant 52, 617796
ASL	100,0%	100,0%	Argininosuccinic aciduria, 207900
ASNS	100,0%	100,0%	Asparagine synthetase deficiency, 615574
ASPA	100,0%	100,0%	Canavan disease, 271900
ASPM	100,0%	100,0%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	100,0%	100,0%	Citrullinemia, 215700
ASXL1	99,9%	99,9%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ASXL2	100,0%	100,0%	Shashi-Pena syndrome, 617190
ASXL3	100,0%	100,0%	Bainbridge-Ropers syndrome, 615485

ATAD1	100,0%	100,0%	Hyperekplexia 4, 618011
ATAD3A	100,0%	100,0%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATIC	100,0%	100,0%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100,0%	100,0%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATN1	100,0%	100,0%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP13A2	100,0%	100,0%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A1	100,0%	100,0%	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 98, 619605 Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	100,0%	100,0%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
ATP2A2	100,0%	100,0%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP1	100,0%	100,0%	Immunodeficiency 47, 300972
ATP6AP2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	100,0%	100,0%	No OMIM Disease ID
ATP6V0A2	100,0%	100,0%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0C	100,0%	100,0%	No OMIM Disease ID
ATP6V1A	100,0%	100,0%	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1B2	100,0%	100,0%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP7A	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400

ATP8A2	100,0%	100,0%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP9A	100,0%	100,0%	No OMIM Disease ID
ATR	100,0%	100,0%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	100,0%	100,0%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Intellectual disability-hypotonic facies syndrome, X-linked, 309580
ATXN2L	100,0%	100,0%	No OMIM Disease ID
AUH	100,0%	100,0%	3-methylglutaconic aciduria, type I, 250950
AUTS2	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVPR2	100,0%	100,0%	Diabetes insipidus, nephrogenic, 1, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B3GALNT2	92,5%	92,5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	99,8%	98,8%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GLCT	100,0%	100,0%	Peters-plus syndrome, 261540
B4GALNT1	100,0%	100,0%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100,0%	100,0%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	96,6%	94,1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100,0%	100,0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAP1	100,0%	100,0%	Tumor predisposition syndrome, 614327 Kury-Isidor syndrome, 619762
BAZ2B	100,0%	100,0%	No OMIM Disease ID
BBS1	100,0%	100,0%	Bardet-Biedl syndrome 1, 209900
BBS10	100,0%	100,0%	Bardet-Biedl syndrome 10, 615987
BBS12	100,0%	100,0%	Bardet-Biedl syndrome 12, 615989
BBS2	100,0%	100,0%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	100,0%	100,0%	Bardet-Biedl syndrome 4, 615982
BBS5	100,0%	100,0%	Bardet-Biedl syndrome 5, 615983
BBS7	100,0%	100,0%	Bardet-Biedl syndrome 7, 615984
BBS9	95,8%	95,8%	Bardet-Biedl syndrome 9, 615986

BCAP31	100,0%	100,0%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	100,0%	100,0%	Hengel-Maroofian-Schols syndrome, 619641
BCKDHA	100,0%	100,0%	Maple syrup urine disease, type Ia, 248600
BCKDHB	100,0%	100,0%	Maple syrup urine disease, type Ib, 248600
BCKDK	100,0%	100,0%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
BCL11A	100,0%	100,0%	Dias-Logan syndrome, 617101
BCL11B	100,0%	99,9%	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 Immunodeficiency 49, 617237
BCOR	100,0%	100,0%	Microphthalmia, syndromic 2, 300166
BCORL1	100,0%	100,0%	Shukla-Vernon syndrome, 301029
BCS1L	100,0%	100,0%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BICRA	100,0%	100,0%	Coffin-Siris syndrome 12, 619325
BLM	100,0%	100,0%	Bloom syndrome, 210900
BLOC1S1	100,0%	100,0%	No OMIM Disease ID
BOLA3	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	100,0%	100,0%	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980
BRAT1	100,0%	100,0%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRF1	100,0%	100,0%	Cerebellofaciodental syndrome, 616202
BRPF1	100,0%	100,0%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	100,0%	100,0%	No OMIM Disease ID
BRWD3	100,0%	100,0%	Intellectual developmental disorder, X-linked 93, 300659
BSCL2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	83,1%	83,1%	Biotinidase deficiency, 253260

BUB1B	100,0%	100,0%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
C12orf4	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 66, 618221
C12orf57	100,0%	100,0%	Temtamy syndrome, 218340
C2CD3	95,9%	95,9%	Orofaciodigital syndrome XIV, 615948
CA2	100,0%	100,0%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	87,7%	87,7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100,0%	100,0%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	100,0%	100,0%	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Developmental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	100,0%	100,0%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	100,0%	100,0%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	100,0%	100,0%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100,0%	100,0%	Developmental and epileptic encephalopathy 69, 618285
CACNA1G	100,0%	100,0%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1I	100,0%	100,0%	No OMIM Disease ID
CACNA2D2	100,0%	100,0%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100,0%	100,0%	Developmental and epileptic encephalopathy 50, 616457
CAMK2A	100,0%	99,9%	Intellectual developmental disorder, autosomal dominant 53, 617798 ?Intellectual developmental disorder, autosomal recessive 63, 618095
CAMK2B	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 54, 617799
CAMK2G	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 59, 618522
CAMK4	100,0%	100,0%	No OMIM Disease ID
CAMTA1	100,0%	100,0%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CANT1	100,0%	100,0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN15	100,0%	100,0%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CARS1	100,0%	100,0%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 27, 616672

CASK	100,0%	100,0%	Intellectual developmental disorder, with or without nystagmus, 300422 Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CBL	100,0%	100,0%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	100,0%	100,0%	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CC2D1A	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 3, 608443
CC2D2A	97,1%	97,1%	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCBE1	100,0%	100,0%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC115	100,0%	100,0%	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	100,0%	100,0%	No OMIM Disease ID
CCDC22	100,0%	100,0%	Ritscher-Schinzel syndrome 2, 300963
CCDC32	100,0%	100,0%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC47	100,0%	100,0%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC88A	97,5%	97,5%	?PEHO syndrome-like, 617507
CCDC88C	100,0%	100,0%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCND2	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	100,0%	100,0%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CDC42	100,0%	100,0%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100,0%	100,0%	No OMIM Disease ID
CDC6	100,0%	100,0%	?Meier-Gorlin syndrome 5, 613805
CDH11	100,0%	100,0%	Teebi hypertelorism syndrome 2, 619736 Elsahy-Waters syndrome, 211380
CDH15	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 3, 612580
CDH2	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDK10	100,0%	100,0%	Al Kaissi syndrome, 617694
CDK13	100,0%	100,0%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	100,0%	100,0%	Developmental and epileptic encephalopathy 87, 618916
CDK5RAP2	100,0%	100,0%	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	100,0%	100,0%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	92,3%	92,2%	Developmental and epileptic encephalopathy 2, 300672

CDKN1C	100,0%	100,0%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDON	100,0%	100,0%	Holoprosencephaly 11, 614226
CELF2	100,0%	100,0%	Developmental and epileptic encephalopathy 97, 619561
CENPF	100,0%	100,0%	Stromme syndrome, 243605
CENPJ	100,0%	100,0%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	100,0%	100,0%	Joubert syndrome 25, 616781
CEP120	100,0%	100,0%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP135	100,0%	100,0%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	100,0%	100,0%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	100,0%	100,0%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	100,0%	100,0%	Joubert syndrome 15, 614464
CEP55	100,0%	100,0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100,0%	100,0%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	100,0%	100,0%	?Seckel syndrome 6, 614728
CEP83	100,0%	100,0%	Nephronophthisis 18, 615862
CEP85L	100,0%	100,0%	Lissencephaly 10, 618873
CEP89	100,0%	100,0%	No OMIM Disease ID
CERT1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 34, 616351
CHAMP1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 40, 616579
CHD1	100,0%	100,0%	Pilarowski-Bjornsson syndrome, 617682
CHD2	100,0%	100,0%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100,0%	100,0%	Snijders Blok-Campeau syndrome, 618205
CHD4	100,0%	100,0%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100,0%	100,0%	No OMIM Disease ID
CHD7	100,0%	100,0%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	100,0%	100,0%	No OMIM Disease ID
CHKB	100,0%	100,0%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100,0%	100,0%	Pontocerebellar hypoplasia, type 8, 614961

CHRM1	100,0%	100,0%	No OMIM Disease ID
CHRNA4	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, 1, 600513
CIC	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 45, 617600
CIT	100,0%	100,0%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	100,0%	100,0%	Filippi syndrome, 272440
CLCN3	96,8%	96,7%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100,0%	100,0%	Raynaud-Claes syndrome, 300114
CLDN11	100,0%	100,0%	Leukodystrophy, hypomyelinating, 22, 619328
CLIC2	100,0%	100,0%	?Intellectual developmental disorder, X-linked, syndromic 32, 300886
CLIP1	100,0%	100,0%	No OMIM Disease ID
CLN3	92,7%	92,5%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	71,7%	71,6%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100,0%	100,0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	100,0%	100,0%	Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLTC	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 56, 617854
CNKSR2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Houge type, 301008
CNNM2	100,0%	100,0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNOT1	100,0%	100,0%	Vissers-Bodmer syndrome, 619033 Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	100,0%	100,0%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100,0%	100,0%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNPY3	100,0%	100,0%	Developmental and epileptic encephalopathy 60, 617929
CNTNAP1	100,0%	100,0%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100,0%	100,0%	Pitt-Hopkins like syndrome 1, 610042
COA8	93,5%	93,5%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100,0%	100,0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COG1	100,0%	100,0%	Congenital disorder of glycosylation, type IIg, 611209

COG4	100,0%	100,0%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	100,0%	100,0%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100,0%	100,0%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type IIl, 614576
COG7	100,0%	100,0%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100,0%	100,0%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	100,0%	100,0%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COL4A2	100,0%	100,0%	Brain small vessel disease 2, 614483
COLEC11	100,0%	100,0%	3MC syndrome 2, 265050
COPB1	100,0%	100,0%	Baralle-Macken syndrome, 619255
COPB2	100,0%	100,0%	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	97,2%	97,2%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX15	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX6B1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
CPE	100,0%	100,0%	BDV syndrome, 619326
CPLANE1	100,0%	100,0%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CPLX1	100,0%	100,0%	Developmental and epileptic encephalopathy 63, 617976
CPS1	100,0%	100,0%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF3	100,0%	100,0%	No OMIM Disease ID
CRADD	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	99,1%	96,1%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREBBP	100,0%	100,0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRLF1	99,6%	98,5%	Cold-induced sweating syndrome 1, 272430
CRPPA	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CSDE1	100,0%	100,0%	No OMIM Disease ID

CSF1R	100,0%	100,0%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSNK1G1	100,0%	100,0%	No OMIM Disease ID
CSNK2A1	94,0%	94,0%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100,0%	100,0%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	100,0%	100,0%	Joubert syndrome 21, 615636
CSTB	100,0%	100,0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	100,0%	99,4%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTDP1	100,0%	100,0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNB1	100,0%	100,0%	Exudative vitreoretinopathy 7, 617572 Pilomatricoma, somatic, 132600 Colorectal cancer, somatic, 114500 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Medulloblastoma, somatic, 155255 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTNND1	100,0%	100,0%	Blepharocheilodontic syndrome 2, 617681
CTNND2	100,0%	100,0%	No OMIM Disease ID
CTSA	100,0%	100,0%	Galactosialidosis, 256540
CTSD	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	100,0%	100,0%	No OMIM Disease ID
CTU2	100,0%	100,0%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUL3	100,0%	100,0%	Neurodevelopmental disorder with or without autism or seizures, 619239 Pseudohypoaldosteronism, type IIE, 614496
CUL4B	100,0%	99,9%	Intellectual developmental disorder, X-linked, syndromic, Cabezas type, 300354
CUX1	100,0%	99,9%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	100,0%	100,0%	Developmental and epileptic encephalopathy 67, 618141
CWC27	100,0%	100,0%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5R3	100,0%	100,0%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYFIP2	100,0%	100,0%	Developmental and epileptic encephalopathy 65, 618008
CYP27A1	100,0%	100,0%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100,0%	100,0%	Spastic paraplegia 56, autosomal recessive, 615030

D2HGDH	100,0%	100,0%	D-2-hydroxyglutaric aciduria, 600721
DAG1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS1	100,0%	100,0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	100,0%	100,0%	Maple syrup urine disease, type II, 248600
DCAF17	100,0%	100,0%	Woodhouse-Sakati syndrome, 241080
DCC	100,0%	100,0%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Esophageal carcinoma, somatic, 133239 Colorectal cancer, somatic, 114500 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DCHS1	100,0%	100,0%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	100,0%	100,0%	Al-Raqad syndrome, 616459
DCX	100,0%	100,0%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDB1	100,0%	100,0%	White-Kernohan syndrome, 619426
DDC	100,0%	100,0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	100,0%	100,0%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	100,0%	100,0%	Warsaw breakage syndrome, 613398
DDX23	100,0%	100,0%	No OMIM Disease ID
DDX3X	99,2%	97,6%	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX59	100,0%	100,0%	Orofaciodigital syndrome V, 174300
DDX6	100,0%	100,0%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	100,0%	100,0%	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEGS1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100,0%	100,0%	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100,0%	100,0%	Epilepsy, familial focal, with variable foci 1, 604364
DHCR24	97,7%	97,7%	Desmosterolosis, 602398
DHCR7	100,0%	100,0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	95,2%	95,2%	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DHFR	100,0%	100,0%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	93,2%	93,2%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480

DHTKD1	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DHX16	100,0%	100,0%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	100,0%	100,0%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX37	100,0%	100,0%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 46, XY sex reversal 11, 273250
DIAPH1	100,0%	100,0%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630
DIS3L2	100,0%	100,0%	Perlman syndrome, 267000
DKC1	100,0%	100,0%	Dyskeratosis congenita, X-linked, 305000
DLAT	100,0%	100,0%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100,0%	100,0%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	100,0%	100,0%	Intellectual developmental disorder, X-linked 90, 300850
DLG4	98,8%	98,8%	Intellectual developmental disorder, autosomal dominant 62, 618793
DLL1	100,0%	100,0%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DMD	100,0%	100,0%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	100,0%	100,0%	Myotonic dystrophy 1, 160900
DMXL2	100,0%	100,0%	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNAJC12	100,0%	100,0%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100,0%	100,0%	3-methylglutaconic aciduria, type V, 610198
DNM1	97,7%	97,4%	Developmental and epileptic encephalopathy 31, 616346
DNM1L	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT3A	100,0%	100,0%	Tatton-Brown-Rahman syndrome, 615879 Acute myeloid leukemia, somatic, 601626 Heyn-Sproul-Jackson syndrome, 618724
DNMT3B	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK3	100,0%	100,0%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	100,0%	100,0%	Adams-Oliver syndrome 2, 614219
DOCK7	100,0%	100,0%	Developmental and epileptic encephalopathy 23, 615859
DOLK	100,0%	100,0%	Congenital disorder of glycosylation, type Im, 610768

DONSON	100,0%	100,0%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	100,0%	100,0%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPF2	100,0%	100,0%	Coffin-Siris syndrome 7, 618027
DPH1	100,0%	100,0%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	99,8%	97,8%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100,0%	100,0%	Congenital disorder of glycosylation, type Iu, 615042
DPP6	100,0%	99,9%	Intellectual developmental disorder, autosomal dominant 33, 616311
DPYD	100,0%	100,0%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100,0%	100,0%	Dihydropyrimidinuria, 222748
DPYSL5	100,0%	100,0%	Ritscher-Schinzel syndrome 4, 619435
DYM	100,0%	100,0%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563
DYNC1I2	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYRK1A	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 7, 614104
EARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	100,0%	100,0%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	100,0%	100,0%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	100,0%	100,0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDC3	100,0%	100,0%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100,0%	100,0%	Congenital disorder of glycosylation, type 2V, 619493
EED	100,0%	99,9%	Cohen-Gibson syndrome, 617561
EEF1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 33, 616409 Intellectual developmental disorder, autosomal dominant 38, 616393
EFNB2	100,0%	100,0%	No OMIM Disease ID
EFTUD2	100,0%	100,0%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	99,9%	99,8%	Kleefstra syndrome 1, 610253
EIF2AK1	100,0%	100,0%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100,0%	100,0%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 Dystonia 33, 619687
EIF2AK3	100,0%	100,0%	Wolcott-Rallison syndrome, 226980

EIF2B4	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B5	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2S3	100,0%	100,0%	MEHMO syndrome, 300148
EIF3F	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A3	100,0%	100,0%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF5A	100,0%	100,0%	Faundes-Banka syndrome, 619376
ELAC2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 17, 615440
ELOVL4	100,0%	100,0%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 58, 617270
EMC1	100,0%	100,0%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EML1	100,0%	100,0%	Band heterotopia, 600348
EMX2	100,0%	100,0%	Schizencephaly, 269160
ENTPD1	100,0%	100,0%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	100,0%	100,0%	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPG5	100,0%	100,0%	Vici syndrome, 242840
EPHA7	100,0%	100,0%	No OMIM Disease ID
ERCC1	100,0%	100,0%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100,0%	100,0%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100,0%	100,0%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC5	100,0%	100,0%	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100,0%	100,0%	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 ?De Sanctis-Cacchione syndrome, 278800 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946

ERCC8	100,0%	100,0%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400
ERLIN2	100,0%	100,0%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	100,0%	100,0%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
ETFB	100,0%	100,0%	Glutaric acidemia IIB, 231680
ETHE1	100,0%	100,0%	Ethylmalonic encephalopathy, 602473
EXOC2	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC7	100,0%	100,0%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOSC2	100,0%	100,0%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100,0%	100,0%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	100,0%	100,0%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100,0%	100,0%	Pontocerebellar hypoplasia, type 1D, 618065
EXTL3	100,0%	100,0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100,0%	100,0%	Weaver syndrome, 277590
FA2H	100,0%	100,0%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	100,0%	100,0%	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	100,0%	100,0%	Joubert syndrome 36, 618763
FAM20C	100,0%	100,0%	Raine syndrome, 259775
FAM50A	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261
FAR1	100,0%	100,0%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	100,0%	100,0%	Rajab interstitial lung disease with brain calcifications 1, 613658
FAT4	100,0%	100,0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBRSL1	100,0%	99,6%	No OMIM Disease ID
FBXL3	100,0%	100,0%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100,0%	100,0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	100,0%	100,0%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	100,0%	100,0%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXW11	100,0%	100,0%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FDFT1	100,0%	100,0%	Squalene synthase deficiency, 618156
FGD1	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	100,0%	100,0%	Developmental and epileptic encephalopathy 47, 617166

FGF13	100,0%	100,0%	Developmental and epileptic encephalopathy 90, 301058
FGF14	100,0%	100,0%	Spinocerebellar ataxia 27, 609307
FGFR1	100,0%	100,0%	Pfeiffer syndrome, 101600 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Hartsfield syndrome, 615465 Trigonocephaly 1, 190440 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	100,0%	100,0%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FGFR3	100,0%	100,0%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 Thanatophoric dysplasia, type II, 187601 Nevus, epidermal, somatic, 162900 CATSHL syndrome, 610474 Thanatophoric dysplasia, type I, 187600 Spermatocytic seminoma, somatic, 273300 Bladder cancer, somatic, 109800 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247

FH	100,0%	100,0%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FIBP	100,0%	100,0%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	100,0%	100,0%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FIGN	100,0%	100,0%	No OMIM Disease ID
FKRP	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615
FLNA	100,0%	100,0%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FLVCR1	100,0%	100,0%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100,0%	100,0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMR1	100,0%	100,0%	Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624 Premature ovarian failure 1, 311360
FOLR1	100,0%	100,0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	100,0%	100,0%	Rett syndrome, congenital variant, 613454
FOXJ1	100,0%	100,0%	Ciliary dyskinesia, primary, 43, 618699
FOXP1	100,0%	100,0%	Intellectual developmental disorder with language impairment with or without autistic features, 613670
FOXP2	100,0%	100,0%	Speech-language disorder-1, 602081
FOXRED1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	100,0%	100,0%	Fraser syndrome 1, 219000

FRMD4A	96,6%	96,6%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMPD4	98,3%	98,3%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100,0%	100,0%	Developmental and epileptic encephalopathy 37, 616981
FTCD	100,0%	100,0%	Glutamate formiminotransferase deficiency, 229100
FTO	94,2%	94,2%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	100,0%	100,0%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100,0%	100,0%	Fucosidosis, 230000
FUT8	100,0%	100,0%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FZR1	100,0%	100,0%	No OMIM Disease ID
GABBR2	100,0%	99,8%	Developmental and epileptic encephalopathy 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903
GABRA1	100,0%	100,0%	Developmental and epileptic encephalopathy 19, 615744
GABRA2	100,0%	100,0%	Developmental and epileptic encephalopathy 78, 618557
GABRA3	100,0%	99,8%	No OMIM Disease ID
GABRA5	100,0%	100,0%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100,0%	100,0%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100,0%	100,0%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	100,0%	100,0%	Developmental and epileptic encephalopathy 43, 617113
GABRD	100,0%	100,0%	No OMIM Disease ID
GABRG2	93,0%	93,0%	Developmental and epileptic encephalopathy 74, 618396 Febrile seizures, familial, 8, 607681 Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100,0%	100,0%	Developmental and epileptic encephalopathy 89, 619124
GALC	100,0%	100,0%	Krabbe disease, 245200
GALE	100,0%	100,0%	Galactose epimerase deficiency, 230350
GALNT2	100,0%	100,0%	Congenital disorder of glycosylation, type IIt, 618885
GALT	100,0%	100,0%	Galactosemia, 230400
GAMT	100,0%	100,0%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	100,0%	100,0%	GAND syndrome, 615074
GATM	100,0%	100,0%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GCH1	100,0%	100,0%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	100,0%	100,0%	?Glycine encephalopathy, 605899
GDI1	100,0%	100,0%	Intellectual developmental disorder, X-linked 41, 300849
GEMIN5	100,0%	100,0%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFAP	100,0%	100,0%	Alexander disease, 203450

GFER	100,0%	100,0%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 39, 618397
GIGYF1	100,0%	100,0%	No OMIM Disease ID
GJA1	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJB1	100,0%	100,0%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	99,9%	99,5%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GK	100,0%	100,0%	Glycerol kinase deficiency, 307030
GLB1	100,0%	100,0%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	100,0%	100,0%	Glycine encephalopathy, 605899
GLI2	100,0%	100,0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100,0%	100,0%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS3	100,0%	100,0%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLS	100,0%	100,0%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Developmental and epileptic encephalopathy 71, 618328
GLUD1	100,0%	100,0%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100,0%	100,0%	Glutamine deficiency, congenital, 610015
GLYCTK	100,0%	100,0%	D-glyceric aciduria, 220120
GM2A	100,0%	100,0%	GM2-gangliosidosis, AB variant, 272750
GMNN	100,0%	100,0%	Meier-Gorlin syndrome 6, 616835
GMPPA	100,0%	100,0%	Alacrima, achalasia, and mental retardation syndrome, 615510

GMPPB	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNAI1	100,0%	100,0%	No OMIM Disease ID
GNAO1	100,0%	100,0%	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAS	83,9%	82,0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ia, 103580 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 McCune-Albright syndrome, somatic, mosaic, 174800 Pseudopseudohypoparathyroidism, 612463
GNB1	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286 Leukemia, acute lymphoblastic, somatic, 613065 Intellectual developmental disorder, autosomal dominant 42, 616973
GNB5	100,0%	100,0%	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 Intellectual developmental disorder with cardiac arrhythmia, 617173
GNPAT	100,0%	100,0%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100,0%	100,0%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	100,0%	100,0%	Mucolipidosis III gamma, 252605
GNS	100,0%	100,0%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	100,0%	100,0%	No OMIM Disease ID
GOT2	100,0%	100,0%	Developmental and epileptic encephalopathy 82, 618721
GPAA1	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	100,0%	99,9%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100,0%	100,0%	Keipert syndrome, 301026
GPHN	100,0%	100,0%	Molybdenum cofactor deficiency C, 615501
GPSM2	100,0%	100,0%	Chudley-McCullough syndrome, 604213
GPT2	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GRIA2	100,0%	100,0%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	99,9%	99,7%	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIA4	100,0%	100,0%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 18, 616204

GRIK2	96,3%	96,3%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 Intellectual developmental disorder, autosomal recessive 6, 611092
GRIN1	100,0%	100,0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Developmental and epileptic encephalopathy 101, 619814 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100,0%	100,0%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100,0%	100,0%	Developmental and epileptic encephalopathy 27, 616139 Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99,9%	99,3%	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100,0%	100,0%	Fraser syndrome 3, 617667
GRM1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 13, 614831 Spinocerebellar ataxia 44, 617691
GRM7	100,0%	100,0%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922
GRN	100,0%	100,0%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GSE1	100,0%	100,0%	No OMIM Disease ID
GSS	100,0%	100,0%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GTF2E2	100,0%	100,0%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	72,5%	72,5%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100,0%	100,0%	Jaberi-Elahi syndrome, 617988
GTPBP3	100,0%	100,0%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	100,0%	100,0%	Mucopolysaccharidosis VII, 253220
H1-4	100,0%	100,0%	Rahman syndrome, 617537
H4C3	100,0%	100,0%	Tessadori-van Haaften neurodevelopmental syndrome 1, 619758
HACE1	100,0%	100,0%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	100,0%	100,0%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	100,0%	100,0%	Trifunctional protein deficiency, 609015
HAX1	100,0%	100,0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	100,0%	100,0%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541

HCN1	98,5%	98,5%	Developmental and epileptic encephalopathy 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482
HDAC4	100,0%	100,0%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC6	100,0%	100,0%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	96,6%	96,0%	Cornelia de Lange syndrome 5, 300882
HEATR5B	100,0%	100,0%	No OMIM Disease ID
HECW2	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	100,0%	100,0%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	100,0%	100,0%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 38, 615516
HESX1	100,0%	100,0%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	100,0%	100,0%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	100,0%	100,0%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	92,1%	92,1%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	100,0%	100,0%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HID1	100,0%	100,0%	No OMIM Disease ID
HIVEP2	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 43, 616977
HK1	100,0%	100,0%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	100,0%	100,0%	Holocarboxylase synthetase deficiency, 253270
HMGCL	100,0%	100,0%	HMG-CoA lyase deficiency, 246450
HNMT	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 51, 616739
HNRNPD	100,0%	100,0%	No OMIM Disease ID
HNRNPH1	100,0%	100,0%	No OMIM Disease ID
HNRNPH2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Bain type, 300986
HNRNPK	100,0%	100,0%	Au-Kline syndrome, 616580
HNRNPU	100,0%	100,0%	Developmental and epileptic encephalopathy 54, 617391
HOXA1	100,0%	100,0%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536

HPD	100,0%	100,0%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPDL	100,0%	100,0%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	100,0%	100,0%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HRAS	100,0%	100,0%	Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Congenital myopathy with excess of muscle spindles, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HS2ST1	100,0%	99,9%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HSD17B10	100,0%	100,0%	HSD10 mitochondrial disease, 300438
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPA9	100,0%	100,0%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	100,0%	100,0%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100,0%	100,0%	3-methylglutaconic aciduria, type VIII, 617248
HUWE1	100,0%	100,0%	Intellectual developmental disorder, X-linked, Turner type, 309590
HYLS1	100,0%	100,0%	Hydrolethalus syndrome, 236680
IARS1	100,0%	100,0%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100,0%	100,0%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
IDS	100,0%	100,0%	Mucopolysaccharidosis II, 309900
IDUA	100,0%	100,0%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IER3IP1	100,0%	100,0%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100,0%	100,0%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250

IFT172	100,0%	100,0%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100,0%	100,0%	Bardet-Biedl syndrome 19, 615996
IFT74	100,0%	100,0%	Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582 ?Bardet-Biedl syndrome 22, 617119
IFT81	95,0%	95,0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	100,0%	100,0%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100,0%	100,0%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100,0%	100,0%	Insulin-like growth factor I, resistance to, 270450
IKBKG	100,0%	100,0%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IL1RAPL1	100,0%	100,0%	Intellectual developmental disorder, X-linked 21, 300143
IMPA1	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 59, 617323
INPP5E	100,0%	100,0%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	100,0%	100,0%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INTS1	100,0%	100,0%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
IPO8	100,0%	100,0%	VISS syndrome, 619472
IQSEC1	100,0%	99,6%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	99,8%	99,1%	Intellectual developmental disorder, X-linked 1, 309530
IREB2	100,0%	100,0%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF2BPL	100,0%	100,0%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRX5	100,0%	100,0%	Hamamy syndrome, 611174
ISCA2	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITGA7	100,0%	100,0%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	100,0%	100,0%	Developmental and epileptic encephalopathy 35, 616647
ITPR1	100,0%	100,0%	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
IVD	100,0%	100,0%	Isovaleric acidemia, 243500
JAG1	100,0%	100,0%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500

JAG2	100,0%	99,6%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAM3	100,0%	100,0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JARID2	100,0%	100,0%	No OMIM Disease ID
JMJD1C	100,0%	100,0%	No OMIM Disease ID
KANK1	100,0%	100,0%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	100,0%	100,0%	Koolen-De Vries syndrome, 610443
KAT5	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	100,0%	100,0%	Arboleda-Tham syndrome, 616268
KAT6B	100,0%	100,0%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KAT8	100,0%	100,0%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100,0%	100,0%	Lissencephaly 6, with microcephaly, 616212
KCNA2	100,0%	100,0%	Developmental and epileptic encephalopathy 32, 616366
KCNA4	100,0%	100,0%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNB1	100,0%	100,0%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100,0%	100,0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	99,8%	98,8%	Spinocerebellar ataxia 13, 605259
KCNH1	98,7%	98,7%	Zimmermann-Laband syndrome 1, 135500 Temple-Baraitser syndrome, 611816
KCNJ10	100,0%	100,0%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100,0%	100,0%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ6	100,0%	100,0%	Keppen-Lubinsky syndrome, 614098
KCNK4	100,0%	100,0%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	97,3%	97,3%	Birk-Barel syndrome, 612292
KCNMA1	100,0%	100,0%	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729
KCNN2	100,0%	100,0%	?Dystonia 34, myoclonic, 619724 Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725
KCNN3	100,0%	100,0%	Zimmermann-Laband syndrome 3, 618658
KCNQ2	100,0%	100,0%	Developmental and epileptic encephalopathy 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200

KCNQ3	100,0%	100,0%	Seizures, benign neonatal, 2, 121201
KCNQ5	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 46, 617601
KCNT1	99,9%	99,6%	Developmental and epileptic encephalopathy 14, 614959 Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100,0%	100,0%	Developmental and epileptic encephalopathy 57, 617771
KCTD3	100,0%	100,0%	No OMIM Disease ID
KCTD7	100,0%	100,0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	100,0%	100,0%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	100,0%	100,0%	Diets-Jongmans syndrome, 618846
KDM4B	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 65, 619320
KDM5B	95,6%	94,1%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDM5C	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6A	100,0%	100,0%	Kabuki syndrome 2, 300867
KDM6B	100,0%	100,0%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KIAA0586	95,8%	95,8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA1109	100,0%	100,0%	Alkuraya-Kucinskas syndrome, 617822
KIDINS220	100,0%	100,0%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 Ventriculomegaly and arthrogryposis, 619501
KIF11	100,0%	100,0%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	100,0%	100,0%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	98,0%	98,0%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF21B	100,0%	100,0%	No OMIM Disease ID
KIF2A	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100,0%	100,0%	Retinitis pigmentosa 89, 618955
KIF4A	100,0%	100,0%	?Intellectual developmental disorder, X-linked 100, 300923
KIF5A	100,0%	100,0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	99,8%	99,8%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100,0%	100,0%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131

KIFBP	96,1%	96,1%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL3	100,0%	100,0%	No OMIM Disease ID
KLF7	100,0%	100,0%	No OMIM Disease ID
KLHL15	100,0%	100,0%	Intellectual developmental disorder, X-linked 103, 300982
KMT2A	100,0%	100,0%	Wiedemann-Steiner syndrome, 605130
KMT2B	99,7%	99,3%	Dystonia 28, childhood-onset, 617284
KMT2C	100,0%	100,0%	Kleefstra syndrome 2, 617768
KMT2D	100,0%	100,0%	Kabuki syndrome 1, 147920
KMT2E	100,0%	100,0%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 51, 617788
KNL1	98,9%	98,9%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 41, 615637
KRAS	100,0%	100,0%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
L1CAM	100,0%	100,0%	MASA syndrome, 303350 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus due to aqueductal stenosis, 307000
L2HGDH	100,0%	100,0%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	100,0%	100,0%	Poretti-Boltshauser syndrome, 615960
LAMA2	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMB1	100,0%	100,0%	Lissencephaly 5, 615191
LAMB2	100,0%	100,0%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMC3	100,0%	100,0%	Cortical malformations, occipital, 614115

LAMP2	100,0%	100,0%	Danon disease, 300257
LARGE1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	100,0%	100,0%	Alazami syndrome, 615071
LARS1	100,0%	100,0%	?Infantile liver failure syndrome 1, 615438
LAS1L	100,0%	100,0%	Wilson-Turner syndrome, 309585
LIAS	100,0%	100,0%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	100,0%	100,0%	LIG4 syndrome, 606593
LINGO1	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 64, 618103
LINS1	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 27, 614340
LMAN2L	100,0%	100,0%	?Intellectual developmental disorder, autosomal recessive 52, 616887
LMBRD2	100,0%	100,0%	Developmental delay with variable neurologic and brain abnormalities, 619694
LMNB1	100,0%	100,0%	Leukodystrophy, adult-onset, autosomal dominant, 169500 Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	100,0%	99,8%	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LONP1	100,0%	100,0%	CODAS syndrome, 600373
LRP2	100,0%	100,0%	Donnai-Barrow syndrome, 222448
LRPPRC	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LSS	100,0%	100,0%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-intellectual disability syndrome 4, 618840
LYRM7	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	100,0%	100,0%	Chediak-Higashi syndrome, 214500
LZTFL1	100,0%	100,0%	Bardet-Biedl syndrome 17, 615994
LZTR1	100,0%	100,0%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAB21L1	100,0%	100,0%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100,0%	100,0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100,0%	100,0%	Lissencephaly 9 with complex brainstem malformation, 618325
MADD	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 DEEAH syndrome, 619004
MAF	94,5%	90,7%	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088
MAG	100,0%	100,0%	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	100,0%	100,0%	Schaaf-Yang syndrome, 615547
MAN1B1	100,0%	100,0%	Rafiq syndrome, 614202

MAN2B1	100,0%	100,0%	Mannosidosis, alpha-, types I and II, 248500
MAN2C1	100,0%	100,0%	Congenital disorder of deglycosylation 2, 619775
MANBA	100,0%	100,0%	Mannosidosis, beta, 248510
MAOA	99,9%	99,4%	Brunner syndrome, 300615
MAP1B	100,0%	100,0%	?Deafness, autosomal dominant 83, 619808 Periventricular nodular heterotopia 9, 618918
MAP2K1	100,0%	100,0%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100,0%	100,0%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100,0%	100,0%	Noonan syndrome 13, 619087
MAPK8IP3	100,0%	100,0%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK5	100,0%	100,0%	No OMIM Disease ID
MAPRE2	100,0%	100,0%	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	100,0%	100,0%	3MC syndrome 1, 257920
MAST1	100,0%	100,0%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAT1A	100,0%	100,0%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 1, 156200
MBOAT7	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS2	100,0%	100,0%	Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 IFAP syndrome with or without BRESHECK syndrome, 308205 ?Olmsted syndrome, X-linked, 300918
MCCC1	100,0%	100,0%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	100,0%	100,0%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	100,0%	100,0%	Mucolipidosis IV, 252650
MCPH1	100,0%	100,0%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	100,0%	100,0%	Developmental and epileptic encephalopathy 51, 617339
MECP2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Encephalopathy, neonatal severe, 300673 Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	100,0%	100,0%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	100,0%	100,0%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895

			Hardikar syndrome, 301068 Opitz-Kaveggia syndrome, 305450
MED12L	100,0%	100,0%	Nizon-Isidor syndrome, 618872
MED13	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 61, 618009
MED13L	100,0%	100,0%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MED17	100,0%	100,0%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	100,0%	100,0%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MED27	84,7%	84,7%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEF2C	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEGF8	100,0%	100,0%	Carpenter syndrome 2, 614976
MEIS2	100,0%	100,0%	Cleft palate, cardiac defects, and mental retardation, 600987
METTL23	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100,0%	99,5%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	100,0%	100,0%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	100,0%	100,0%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100,0%	100,0%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100,0%	100,0%	Congenital disorder of glycosylation, type IIa, 212066
MGP	100,0%	100,0%	Keutel syndrome, 245150
MIA3	100,0%	100,0%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MICU1	100,0%	100,0%	Myopathy with extrapyramidal signs, 615673
MID1	100,0%	100,0%	Opitz GBBB syndrome, 300000
MID2	100,0%	100,0%	?Intellectual developmental disorder, X-linked 101, 300928
MINPP1	100,0%	100,0%	Pontocerebellar hypoplasia, type 16, 619527
MKKS	100,0%	100,0%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	100,0%	100,0%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MLC1	100,0%	100,0%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	100,0%	100,0%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100,0%	100,0%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100,0%	100,0%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400

MMADHC	89,7%	89,7%	Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Homocystinuria, cblD type, variant 1, 277410
MMGT1	100,0%	100,0%	No OMIM Disease ID
MMUT	100,0%	100,0%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100,0%	100,0%	CEBALID syndrome, 618774 Meningioma, 607174
MOCS1	100,0%	100,0%	Molybdenum cofactor deficiency A, 252150
MOCS2	100,0%	100,0%	Molybdenum cofactor deficiency B, 252160
MOGS	100,0%	100,0%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPDU1	100,0%	100,0%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100,0%	100,0%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPLKIP	100,0%	100,0%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPV17	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRAS	100,0%	100,0%	Noonan syndrome 11, 618499
MRPS22	100,0%	100,0%	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719
MRPS34	100,0%	100,0%	Combined oxidative phosphorylation deficiency 32, 617664
MSL2	100,0%	100,0%	No OMIM Disease ID
MSL3	98,4%	97,1%	Basilicata-Akhtar syndrome, 301032
MSMO1	100,0%	100,0%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTFMT	100,0%	100,0%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	100,0%	100,0%	Homocystinuria due to MTHFR deficiency, 236250
MTHFS	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTO1	94,3%	92,1%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTR	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
C12orf65	100,0%	100,0%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTRR	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270

MVK	90,5%	90,5%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYCN	100,0%	100,0%	Feingold syndrome 1, 164280
MYH9	100,0%	100,0%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYO5A	100,0%	100,0%	Griscelli syndrome, type 1, 214450
MYO9A	100,0%	100,0%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYT1L	90,2%	90,2%	Intellectual developmental disorder, autosomal dominant 39, 616521
NAA10	100,0%	100,0%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	96,8%	96,8%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 73, 619717
NACC1	100,0%	100,0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAGA	100,0%	100,0%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	99,8%	99,8%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS1	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAXE	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	100,0%	100,0%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBN	100,0%	100,0%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCAPG2	100,0%	100,0%	Khan-Khan-Katsanis syndrome, 618460
NCDN	100,0%	100,0%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCKAP1	100,0%	100,0%	No OMIM Disease ID
NDE1	100,0%	100,0%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013

NDP	100,0%	100,0%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFAF3	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	95,3%	91,3%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	100,0%	100,0%	Periventricular nodular heterotopia 7, 617201
NEMF	100,0%	100,0%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEU1	100,0%	100,0%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROD2	100,0%	100,0%	Developmental and epileptic encephalopathy 72, 618374
NEXMIF	100,0%	100,0%	Intellectual developmental disorder, X-linked 98, 300912
NF1	100,0%	100,0%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFE2L2	100,0%	100,0%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	99,2%	99,2%	Brain malformations with or without urinary tract defects, 613735
NFIB	100,0%	100,0%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100,0%	99,7%	Marshall-Smith syndrome, 602535 Malan syndrome, 614753
NFU1	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 1, 605711

NGLY1	100,0%	100,0%	Congenital disorder of deglycosylation 1, 615273
NHLRC2	100,0%	100,0%	FINCA syndrome, 618278
NHS	100,0%	100,0%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIPBL	100,0%	100,0%	Cornelia de Lange syndrome 1, 122470
NKAP	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
NKX2-1	100,0%	100,0%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN2	100,0%	100,0%	No OMIM Disease ID
NLGN3	100,0%	100,0%	No OMIM Disease ID
NLGN4X	100,0%	100,0%	Intellectual developmental disorder, X-linked, 300495
NONO	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOVA2	100,0%	100,0%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPC1	100,0%	100,0%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100,0%	100,0%	Niemann-pick disease, type C2, 607625
NPHP1	100,0%	100,0%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NR2F1	100,0%	99,8%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	100,0%	100,0%	No OMIM Disease ID
NRAS	100,0%	100,0%	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
NRCAM	100,0%	100,0%	No OMIM Disease ID
NRROS	100,0%	100,0%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100,0%	100,0%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	100,0%	100,0%	Sotos syndrome, 117550
NSD2	100,0%	100,0%	Rauch-Steindl syndrome, 619695
NSDHL	100,0%	100,0%	CK syndrome, 300831 CHILD syndrome, 308050
NSF	100,0%	100,0%	Developmental and epileptic encephalopathy 96, 619340

NSUN2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 5, 611091
NT5C2	100,0%	100,0%	Spastic paraplegia 45, autosomal recessive, 613162
NTNG2	100,0%	100,0%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100,0%	100,0%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	100,0%	100,0%	Developmental and epileptic encephalopathy 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUBPL	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDT2	100,0%	100,0%	No OMIM Disease ID
NUP107	100,0%	100,0%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP188	100,0%	100,0%	Sandestig-Stefanova syndrome, 618804
NUP214	100,0%	100,0%	Leukemia, T-cell acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, somatic, 601626
NUP62	100,0%	100,0%	Striatonigral degeneration, infantile, 271930
NUP85	100,0%	100,0%	Nephrotic syndrome, type 17, 618176
NUS1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	100,0%	100,0%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	100,0%	100,0%	Pseudo-TORCH syndrome 1, 251290
OCRL	100,0%	100,0%	Dent disease 2, 300555 Lowe syndrome, 309000
ODC1	100,0%	100,0%	Bachmann-Bupp syndrome, 619075
OFD1	100,0%	100,0%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OGDHL	100,0%	100,0%	Yoon-Bellen neurodevelopmental syndrome, 619701
OGT	100,0%	100,0%	Intellectual developmental disorder, X-linked 106, 300997
OPA3	100,0%	100,0%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	100,0%	99,5%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
ORC1	100,0%	100,0%	Meier-Gorlin syndrome 1, 224690
OSGEP	100,0%	100,0%	Galloway-Mowat syndrome 3, 617729
OTC	100,0%	100,0%	Ornithine transcarbamylase deficiency, 311250
OTUD5	100,0%	99,7%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	100,0%	100,0%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452

OTUD7A	99,7%	98,7%	No OMIM Disease ID
OTX2	100,0%	100,0%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125
OXR1	100,0%	100,0%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P4HTM	100,0%	100,0%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PACS1	100,0%	100,0%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100,0%	100,0%	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	100,0%	100,0%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	100,0%	100,0%	Phenylketonuria, 261600
PAK1	100,0%	100,0%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	100,0%	100,0%	Intellectual developmental disorder, X-linked 30, 300558
MPP5	100,0%	100,0%	No OMIM Disease ID
PAM16	82,9%	82,9%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	100,0%	100,0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	100,0%	100,0%	Oocyte maturation defect 7, 618550
PARN	89,5%	87,8%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARP6	100,0%	100,0%	No OMIM Disease ID
PAX1	100,0%	100,0%	Otofaciocervical syndrome 2, 615560
PAX6	100,0%	100,0%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200 ?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190
PAX7	100,0%	100,0%	Rhabdomyosarcoma 2, alveolar, 268220 Myopathy, congenital, progressive, with scoliosis, 618578
PAX8	100,0%	100,0%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	100,0%	100,0%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	100,0%	100,0%	Pyruvate carboxylase deficiency, 266150

PCCA	100,0%	100,0%	Propionicacidemia, 606054
PCCB	99,9%	98,1%	Propionicacidemia, 606054
PCDH12	100,0%	100,0%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH19	100,0%	100,0%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	100,0%	100,0%	No OMIM Disease ID
PCGF2	100,0%	100,0%	Turnpenny-Fry syndrome, 618371
PCLO	100,0%	100,0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	100,0%	100,0%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT2	100,0%	100,0%	Spastic paraplegia 82, autosomal recessive, 618770
PDE2A	100,0%	100,0%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE4D	100,0%	100,0%	Acrodysostosis 2, with or without hormone resistance, 614613
PDGFRB	100,0%	100,0%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007
PDHA1	100,0%	100,0%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100,0%	100,0%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100,0%	100,0%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100,0%	100,0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	97,4%	97,4%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	100,0%	100,0%	Prolidase deficiency, 170100
PET100	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	100,0%	100,0%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100,0%	100,0%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100,0%	100,0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100,0%	100,0%	Peroxisome biogenesis disorder 12A (Zellweger), 614886

PEX2	100,0%	100,0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100,0%	100,0%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100,0%	100,0%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	100,0%	100,0%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100,0%	100,0%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGAP1	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	100,0%	100,0%	Phosphoglycerate kinase 1 deficiency, 300653
PGM2L1	100,0%	100,0%	No OMIM Disease ID
PGM3	91,7%	91,7%	Immunodeficiency 23, 615816
PHACTR1	100,0%	100,0%	Developmental and epileptic encephalopathy 70, 618298
PHF21A	100,0%	100,0%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF6	100,0%	100,0%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263
PHGDH	100,0%	100,0%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	100,0%	99,9%	Chung-Jansen syndrome, 617991
PI4KA	100,0%	99,9%	Spastic paraplegia 84, autosomal recessive, 619621 Gastrointestinal defects and immunodeficiency syndrome 2, 619708 Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	100,0%	100,0%	Joubert syndrome 33, 617767
PIDD1	100,0%	100,0%	No OMIM Disease ID
PIGA	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100,0%	100,0%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816

PIGF	100,0%	100,0%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356
PIGG	100,0%	100,0%	Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	80,3%	74,7%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100,0%	100,0%	CHIME syndrome, 280000
PIGN	98,8%	98,8%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	100,0%	100,0%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100,0%	100,0%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	100,0%	100,0%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100,0%	100,0%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100,0%	99,9%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	100,0%	100,0%	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108 Macrodactyly, somatic,,
PIK3R2	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PISD	100,0%	100,0%	Liberfarb syndrome, 618889
PITRM1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PJA1	100,0%	100,0%	No OMIM Disease ID
PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLAA	100,0%	100,0%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527

PLCB1	100,0%	100,0%	Developmental and epileptic encephalopathy 12, 613722
PLK1	100,0%	100,0%	No OMIM Disease ID
PLK4	100,0%	100,0%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	100,0%	100,0%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	100,0%	100,0%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLXNA1	100,0%	100,0%	No OMIM Disease ID
PLXNA2	100,0%	100,0%	No OMIM Disease ID
PLXND1	100,0%	100,0%	No OMIM Disease ID
PMM2	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	100,0%	100,0%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	100,0%	100,0%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	100,0%	100,0%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
POGZ	100,0%	100,0%	White-Sutton syndrome, 616364
POLA1	100,0%	100,0%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLG	100,0%	100,0%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR1C	83,0%	82,8%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR2A	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100,0%	100,0%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POMGNT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157

			Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	100,0%	100,0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
PORCN	100,0%	100,0%	Focal dermal hypoplasia, 305600
POU1F1	100,0%	100,0%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU3F3	99,9%	99,0%	Snijders Blok-Fisher syndrome, 618604
PPIL1	100,0%	100,0%	Pontocerebellar hypoplasia, type 14, 619301
PPM1D	100,0%	100,0%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	100,0%	100,0%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	100,0%	100,0%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	100,0%	100,0%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PPP2CA	100,0%	100,0%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	93,6%	93,6%	Intellectual developmental disorder, autosomal dominant 36, 616362
PPP2R5B	100,0%	100,0%	No OMIM Disease ID
PPP2R5C	100,0%	100,0%	No OMIM Disease ID
PPP2R5D	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 35, 616355
PPP3CA	100,0%	100,0%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Developmental and epileptic encephalopathy 91, 617711
PPT1	82,5%	82,5%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100,0%	100,0%	Renpenning syndrome, 309500
PRDM15	100,0%	99,9%	No OMIM Disease ID
PRICKLE2	100,0%	100,0%	No OMIM Disease ID
PRKACB	100,0%	100,0%	Cardioacrofacial dysplasia 2, 619143
PRKAR1A	100,0%	100,0%	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Carney complex, type 1, 160980

			Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,
PRKAR1B	100,0%	100,0%	Marbach-Schaaf neurodevelopmental syndrome, 619680
PRMT7	100,0%	100,0%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	100,0%	100,0%	Hyperprolinemia, type I, 239500
PRPS1	100,0%	100,0%	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PRR12	100,0%	100,0%	Neuroocular syndrome, 619539
PRSS12	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 1, 249500
PRUNE1	93,6%	93,6%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PSAP	100,0%	100,0%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PSAT1	100,0%	100,0%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMC5	100,0%	100,0%	No OMIM Disease ID
PSMD12	100,0%	100,0%	Stankiewicz-Isidor syndrome, 617516
PSPH	100,0%	100,0%	Phosphoserine phosphatase deficiency, 614023
PTCH1	100,0%	100,0%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTCHD1	100,0%	100,0%	No OMIM Disease ID
PTDSS1	100,0%	100,0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	100,0%	100,0%	Cowden syndrome 1, 158350 Lhermitte-Duclos disease, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTF1A	100,0%	100,0%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTPN11	100,0%	100,0%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN23	100,0%	100,0%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890

PTRH2	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100,0%	100,0%	No OMIM Disease ID
PTS	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100,0%	100,0%	Verheij syndrome, 615583
PUM1	100,0%	100,0%	Spinocerebellar ataxia 47, 617931
PURA	100,0%	100,0%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	100,0%	99,2%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100,0%	100,0%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PYCR1	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100,0%	100,0%	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100,0%	100,0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100,0%	100,0%	Ververi-Brady syndrome, 617982
RAB11B	100,0%	100,0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	100,0%	100,0%	No OMIM Disease ID
RAB18	100,0%	100,0%	Warburg micro syndrome 3, 614222
RAB23	100,0%	100,0%	Carpenter syndrome, 201000
RAB27A	100,0%	100,0%	Griscelli syndrome, type 2, 607624
RAB39B	100,0%	100,0%	Intellectual developmental disorder, X-linked 72, 300271 Waisman syndrome, 311510
RAB3GAP1	99,4%	99,4%	Martsolf syndrome 2, 619420 Warburg micro syndrome 1, 600118
RAB3GAP2	100,0%	100,0%	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225
RAC1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAC3	100,0%	100,0%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	100,0%	100,0%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RAF1	100,0%	100,0%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAI1	100,0%	100,0%	Smith-Magenis syndrome, 182290
RALA	100,0%	100,0%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RARB	100,0%	100,0%	Microphthalmia, syndromic 12, 615524

RARS1	94,4%	94,4%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100,0%	100,0%	Pontocerebellar hypoplasia, type 6, 611523
RBBP8	100,0%	100,0%	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBFOX1	100,0%	99,7%	No OMIM Disease ID
RBM10	100,0%	100,0%	TARP syndrome, 311900
RBM28	100,0%	100,0%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	100,0%	100,0%	Adams-Oliver syndrome 3, 614814
RCBTB1	100,0%	100,0%	Retinal dystrophy with or without extraocular anomalies, 617175
RECQL4	100,0%	100,0%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
RELN	100,0%	100,0%	Lissencephaly 2 (Norman-Roberts type), 257320
RERE	99,9%	99,9%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	97,8%	97,6%	No OMIM Disease ID
RFT1	100,0%	100,0%	Congenital disorder of glycosylation, type In, 612015
RFX3	100,0%	100,0%	No OMIM Disease ID
RFX4	100,0%	100,0%	No OMIM Disease ID
RFX7	99,1%	99,1%	No OMIM Disease ID
RHEB	100,0%	100,0%	No OMIM Disease ID
RHOBTB2	100,0%	100,0%	Developmental and epileptic encephalopathy 64, 618004
RIC1	100,0%	100,0%	CATIFA syndrome, 618761
RIMS2	97,8%	97,8%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIT1	100,0%	100,0%	Noonan syndrome 8, 615355
RLIM	100,0%	100,0%	Tonne-Kalscheuer syndrome, 300978
RMND1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH2A	100,0%	100,0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91,0%	91,0%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0%	100,0%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	100,0%	100,0%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100,0%	100,0%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	100,0%	100,0%	Tenorio syndrome, 616260
RNF13	100,0%	100,0%	Developmental and epileptic encephalopathy 73, 618379

RNF2	100,0%	100,0%	Luo-Schoch-Yamamoto syndrome, 619460
RNF220	100,0%	100,0%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNPC3	100,0%	100,0%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU4ATAC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROGDI	100,0%	100,0%	Kohlschutter-Tonz syndrome, 226750
ROR2	97,0%	97,0%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	100,0%	100,0%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPGRIP1L	100,0%	99,8%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPIA	100,0%	100,0%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, 35, 300998
RPS19	100,0%	100,0%	Diamond-Blackfan anemia 1, 105650
RPS6KA3	100,0%	99,8%	Intellectual developmental disorder, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RRM2B	100,0%	100,0%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RRP7A	100,0%	99,9%	?Microcephaly 28, primary, autosomal recessive, 619453
RSPRY1	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RTN4IP1	100,0%	100,0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	100,0%	100,0%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 61, 617773
RXYLT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SALL1	100,0%	100,0%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480

SAMD9	100,0%	100,0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMHD1	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS1	100,0%	100,0%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SATB1	100,0%	100,0%	Kohlschutter-Tonz syndrome-like, 619229 Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	100,0%	100,0%	Glass syndrome, 612313
SBDS	100,0%	100,0%	Shwachman-Diamond syndrome, 260400
SC5D	100,0%	100,0%	Lathosterolosis, 607330
SCAF4	100,0%	100,0%	No OMIM Disease ID
SCAMP5	100,0%	100,0%	No OMIM Disease ID
SCAPER	100,0%	100,0%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCN1A	100,0%	100,0%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 Migraine, familial hemiplegic, 3, 609634 Dravet syndrome, 607208 Febrile seizures, familial, 3A, 604403 Generalized epilepsy with febrile seizures plus, type 2, 604403
SCN1B	100,0%	100,0%	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN2A	100,0%	100,0%	Seizures, benign familial infantile, 3, 607745 Developmental and epileptic encephalopathy 11, 613721 Episodic ataxia, type 9, 618924
SCN3A	100,0%	100,0%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN8A	100,0%	100,0%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558
SCO1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100,0%	100,0%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCUBE3	100,0%	100,0%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SCYL1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 21, 616719

SDCCAG8	100,0%	100,0%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SDHA	100,0%	100,0%	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SEC31A	100,0%	100,0%	?Halperin-Birk syndrome, 618651
SEMA3E	100,0%	100,0%	?CHARGE syndrome, 214800
SEPSECS	100,0%	100,0%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	100,0%	99,4%	Intellectual developmental disorder, autosomal dominant 58, 618106
SETBP1	100,0%	100,0%	Schinzel-Giedion midface retraction syndrome, 269150 Intellectual developmental disorder, autosomal dominant 29, 616078
SETD1A	100,0%	100,0%	Epilepsy, early-onset, with or without developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	100,0%	100,0%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100,0%	100,0%	Luscan-Lumish syndrome, 616831
SETD5	98,0%	98,0%	Intellectual developmental disorder, autosomal dominant 23, 615761
SFXN4	100,0%	100,0%	Combined oxidative phosphorylation deficiency 18, 615578
SGPL1	100,0%	100,0%	Nephrotic syndrome, type 14, 617575
SGSH	100,0%	100,0%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK2	98,9%	98,9%	No OMIM Disease ID
SHANK3	98,3%	97,3%	Phelan-McDermid syndrome, 606232
SHH	100,0%	100,0%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHMT2	100,0%	100,0%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC2	100,0%	100,0%	Noonan syndrome-like with loose anagen hair 1, 607721
SHROOM4	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434
SIAH1	100,0%	100,0%	Buratti-Harel syndrome, 619314
SIK1	100,0%	100,0%	Developmental and epileptic encephalopathy 30, 616341
SIL1	100,0%	100,0%	Marinesco-Sjogren syndrome, 248800
SIN3A	100,0%	100,0%	Witteveen-Kolk syndrome, 613406
SIN3B	100,0%	100,0%	No OMIM Disease ID
SIX3	100,0%	100,0%	Schizencephaly, 269160 Holoprosencephaly 2, 157170

SKI	100,0%	100,0%	Shprintzen-Goldberg syndrome, 182212
SLC12A2	100,0%	100,0%	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081
SLC12A5	97,4%	97,4%	Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100,0%	100,0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	100,0%	100,0%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A2	100,0%	100,0%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	100,0%	100,0%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	98,7%	98,7%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	100,0%	100,0%	Dicarboxylic aminoaciduria, 222730
SLC1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 41, 617105
SLC1A4	100,0%	100,0%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	100,0%	100,0%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A12	100,0%	100,0%	Developmental and epileptic encephalopathy 39, 612949
SLC25A15	100,0%	100,0%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100,0%	100,0%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99,7%	99,7%	Fontaine progeroid syndrome, 612289
SLC25A42	100,0%	100,0%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC2A1	100,0%	100,0%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC33A1	100,0%	100,0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	100,0%	100,0%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100,0%	100,0%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	81,0%	81,0%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35C1	100,0%	100,0%	Congenital disorder of glycosylation, type IIc, 266265
SLC39A14	93,6%	93,5%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A8	100,0%	100,0%	Congenital disorder of glycosylation, type IIn, 616721
SLC45A1	100,0%	100,0%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC46A1	100,0%	100,0%	Folate malabsorption, hereditary, 229050
SLC4A4	100,0%	100,0%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278

SLC5A6	100,0%	100,0%	Neurodegeneration, infantile-onset, biotin-responsive, 618973
SLC6A1	100,0%	100,0%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	100,0%	100,0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A3	100,0%	100,0%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A8	100,0%	100,0%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100,0%	100,0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100,0%	100,0%	Lysinuric protein intolerance, 222700
SLC9A6	100,0%	99,7%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SLC9A7	100,0%	100,0%	Intellectual developmental disorder, X-linked 108, 301024
SMAD4	100,0%	100,0%	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA1	100,0%	99,9%	No OMIM Disease ID
SMARCA2	98,4%	98,2%	Nicolaides-Baraitser syndrome, 601358 Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100,0%	100,0%	Coffin-Siris syndrome 4, 614609
SMARCA5	100,0%	100,0%	No OMIM Disease ID
SMARCB1	100,0%	100,0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCC2	100,0%	100,0%	Coffin-Siris syndrome 8, 618362
SMARCD1	100,0%	100,0%	Coffin-Siris syndrome 11, 618779
SMARCE1	100,0%	100,0%	Coffin-Siris syndrome 5, 616938
SMC1A	100,0%	100,0%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100,0%	100,0%	Cornelia de Lange syndrome 3, 610759
SMG8	100,0%	100,0%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100,0%	100,0%	Heart and brain malformation syndrome, 616920
SMOC1	100,0%	100,0%	Microphthalmia with limb anomalies, 206920
SMPD1	100,0%	100,0%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SMPD4	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAP25	100,0%	100,0%	?Myasthenic syndrome, congenital, 18, 616330

SNAP29	100,0%	100,0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORD118	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRPB	100,0%	100,0%	Cerebrocostomandibular syndrome, 117650
SNRPN	100,0%	100,0%	Prader-Willi syndrome, 176270
SNX14	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100,0%	100,0%	No OMIM Disease ID
SOBP	100,0%	99,7%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	100,0%	100,0%	ZTTK syndrome, 617140
SOS1	100,0%	100,0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100,0%	100,0%	Noonan syndrome 9, 616559
SOX10	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100,0%	100,0%	Coffin-Siris syndrome 9, 615866
SOX2	100,0%	100,0%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX3	100,0%	100,0%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX4	100,0%	100,0%	Coffin-Siris syndrome 10, 618506
SOX5	100,0%	100,0%	Lamb-Shaffer syndrome, 616803
SOX6	100,0%	100,0%	Tolchin-Le Caignec syndrome, 618971
SPART	100,0%	100,0%	Troyer syndrome, 275900
SPAST	100,0%	100,0%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	100,0%	100,0%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPECC1L	97,8%	96,2%	Teebi hypertelorism syndrome 1, 145420 ?Facial clefting, oblique, 1, 600251
SPEN	100,0%	100,0%	Radio-Tartaglia syndrome, 619312
SPG11	100,0%	100,0%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPOCK1	100,0%	100,0%	No OMIM Disease ID
SPOP	100,0%	100,0%	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829
SPR	100,0%	100,0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100,0%	100,0%	Legius syndrome, 611431

SPRED2	100,0%	100,0%	Noonan syndrome 14, 619745
SPTAN1	100,0%	100,0%	Developmental and epileptic encephalopathy 5, 613477
SPTBN1	100,0%	100,0%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100,0%	99,9%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SRCAP	100,0%	100,0%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140
SRD5A3	100,0%	100,0%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SRP54	100,0%	100,0%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRPX2	100,0%	100,0%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM2	100,0%	100,0%	No OMIM Disease ID
SSR4	100,0%	100,0%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	95,8%	95,2%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98,7%	98,7%	Salt and pepper developmental regression syndrome, 609056
STAG1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 47, 617635
STAG2	99,9%	99,4%	Holoprosencephaly 13, X-linked, 301043 Mullegama-Klein-Martinez syndrome, 301022
STAMBP	100,0%	100,0%	Microcephaly-capillary malformation syndrome, 614261
CXorf56	100,0%	100,0%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	100,0%	100,0%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	100,0%	100,0%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100,0%	100,0%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STT3A	100,0%	100,0%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100,0%	100,0%	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	100,0%	100,0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	100,0%	100,0%	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	100,0%	99,9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100,0%	100,0%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100,0%	100,0%	Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
SUMF1	100,0%	100,0%	Multiple sulfatase deficiency, 272200

SUOX	100,0%	100,0%	Sulfite oxidase deficiency, 272300
SUPT16H	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480
SURF1	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	100,0%	100,0%	Imagawa-Matsumoto syndrome, 618786
SVBP	100,0%	100,0%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYN1	100,0%	100,0%	Intellectual developmental disorder, X-linked 50, 300115 Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	100,0%	100,0%	No OMIM Disease ID
SYNGAP1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100,0%	100,0%	Parkinson disease 20, early-onset, 615530 Developmental and epileptic encephalopathy 53, 617389
SYP	100,0%	100,0%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	100,0%	100,0%	Baker-Gordon syndrome, 618218
SZT2	100,0%	100,0%	Developmental and epileptic encephalopathy 18, 615476
TACO1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAF1	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TAF13	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 60, 617432
TAF1C	100,0%	100,0%	No OMIM Disease ID
TAF2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 40, 615599
TAF6	100,0%	100,0%	Alazami-Yuan syndrome, 617126
TANC2	100,0%	100,0%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100,0%	100,0%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TASP1	100,0%	100,0%	Suleiman-El-Hattab syndrome, 618950
TAT	100,0%	100,0%	Tyrosinemia, type II, 276600
TBC1D20	100,0%	100,0%	Warburg micro syndrome 4, 615663
TBC1D23	100,0%	100,0%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100,0%	100,0%	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal dominant 65, 616044 Developmental and epileptic encephalopathy 16, 615338 DOORS syndrome, 220500
TBC1D2B	99,9%	99,7%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D7	100,0%	100,0%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000

TBCD	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100,0%	100,0%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBP	100,0%	100,0%	Spinocerebellar ataxia 17, 607136
TBR1	100,0%	100,0%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	98,1%	95,9%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TCF20	100,0%	100,0%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF4	100,0%	100,0%	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	100,0%	100,0%	No OMIM Disease ID
TCN2	100,0%	100,0%	Transcobalamin II deficiency, 275350
TCTN2	100,0%	100,0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100,0%	100,0%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDP2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 14, 614020
TELO2	100,0%	100,0%	You-Hoover-Fong syndrome, 616954
TENM3	100,0%	100,0%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TET3	100,0%	100,0%	Beck-Fahrner syndrome, 618798
TFAP2A	100,0%	100,0%	Branchiooculofacial syndrome, 113620
TFE3	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, with pigmentary mosaicism and coarse facies, 301066 Renal cell carcinoma, papillary, 1, 300854
TGDS	100,0%	100,0%	Catel-Manzke syndrome, 616145
TGFBR1	100,0%	99,9%	Loeys-Dietz syndrome 1, 609192
TGIF1	100,0%	100,0%	Holoprosencephaly 4, 142946
TH	100,0%	100,0%	Segawa syndrome, recessive, 605407
THOC2	100,0%	100,0%	Intellectual developmental disorder, X-linked 12, 300957

THOC6	100,0%	100,0%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100,0%	100,0%	Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650
THUMPD1	100,0%	100,0%	No OMIM Disease ID
TIMM50	100,0%	100,0%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100,0%	100,0%	Mohr-Tranebjaerg syndrome, 304700
TINF2	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TKFC	100,0%	100,0%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98,8%	98,7%	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 57, 618050
TMCO1	88,0%	88,0%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM106B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	100,0%	100,0%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	100,0%	100,0%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM222	100,0%	100,0%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	100,0%	100,0%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100,0%	100,0%	Joubert syndrome 14, 614424
TMEM240	100,0%	100,0%	Spinocerebellar ataxia 21, 607454
TMEM63A	100,0%	100,0%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM67	100,0%	100,0%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TMEM70	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100,0%	100,0%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMLHE	99,6%	99,5%	No OMIM Disease ID
TMTC3	100,0%	100,0%	Lissencephaly 8, 617255
TMX2	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNIK	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 54, 617028
TNPO2	100,0%	100,0%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNR	100,0%	100,0%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNRC6B	100,0%	100,0%	Global developmental delay with speech and behavioral abnormalities, 619243

TOE1	100,0%	100,0%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100,0%	100,0%	Joubert syndrome 37, 619185
TOMM70	100,0%	100,0%	No OMIM Disease ID
TOR1A	92,9%	91,5%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TP53RK	100,0%	100,0%	Galloway-Mowat syndrome 4, 617730
TP73	100,0%	100,0%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPI1	100,0%	100,0%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	100,0%	100,0%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100,0%	100,0%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TPRKB	82,3%	81,9%	Galloway-Mowat syndrome 5, 617731
TRAF7	100,0%	100,0%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100,0%	100,0%	Seckel syndrome 9, 616777
TRAK1	100,0%	100,0%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2L	100,0%	100,0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	100,0%	100,0%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 13, 613192
TREX1	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	100,0%	100,0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM8	100,0%	100,0%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	99,9%	99,7%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIP12	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIT1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100,0%	100,0%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	100,0%	100,0%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM3	100,0%	100,0%	No OMIM Disease ID

TRRAP	100,0%	100,0%	?Deafness, autosomal dominant 75, 618778 Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100,0%	100,0%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TSEN15	100,0%	100,0%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100,0%	100,0%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100,0%	100,0%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	94,9%	94,9%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100,0%	100,0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	100,0%	100,0%	Intellectual developmental disorder, X-linked 58, 300210
TTC19	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	100,0%	100,0%	Trichohepatoenteric syndrome 1, 222470
TTC5	100,0%	100,0%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC8	100,0%	100,0%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 39, 615541
TUBA1A	100,0%	100,0%	Lissencephaly 3, 611603
TUBA8	100,0%	100,0%	No OMIM Disease ID
TUBB	100,0%	99,8%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB2A	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	99,5%	97,4%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	97,0%	97,0%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	100,0%	100,0%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100,0%	100,0%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 7, 611093

TWIST1	100,0%	100,0%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWNK	100,0%	100,0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
U2AF2	100,0%	100,0%	No OMIM Disease ID
UBA5	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE2A	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Nascimento type, 300860
UBE3A	100,0%	100,0%	Angelman syndrome, 105830
UBE3B	100,0%	100,0%	Kaufman oculocerebrofacial syndrome, 244450
UBE4A	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639
UBR1	98,0%	98,0%	Johanson-Blizzard syndrome, 243800
UBR7	100,0%	100,0%	Li-Campeau syndrome, 619189
UBTF	100,0%	100,0%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	100,0%	100,0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100,0%	100,0%	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
UGDH	100,0%	100,0%	Developmental and epileptic encephalopathy 84, 618792
UGP2	96,6%	96,3%	Developmental and epileptic encephalopathy 83, 618744
UNC13A	100,0%	100,0%	No OMIM Disease ID
UNC80	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	100,0%	100,0%	Beta-ureidopropionase deficiency, 613161
UPF1	99,6%	99,0%	No OMIM Disease ID
UPF3B	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UROC1	100,0%	100,0%	?Urocanase deficiency, 276880
USP27X	100,0%	100,0%	Intellectual developmental disorder, X-linked 105, 300984
USP7	94,8%	94,8%	Hao-Fountain syndrome, 616863
USP9X	100,0%	100,0%	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
VAMP1	100,0%	100,0%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100,0%	100,0%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VARS1	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802

VARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 20, 615917
VLDLR	100,0%	100,0%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	100,0%	100,0%	?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	99,5%	99,4%	Cohen syndrome, 216550
VPS16	100,0%	100,0%	Dystonia 30, 619291
VPS35L	100,0%	100,0%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	100,0%	100,0%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS4A	100,0%	100,0%	CIMDAG syndrome, 619273
VPS50	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS53	100,0%	99,8%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	100,0%	100,0%	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	100,0%	100,0%	Desanto-Shinawi syndrome, 616708
WARS2	100,0%	100,0%	Parkinsonism-dystonia 3, childhood-onset, 619738 Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	100,0%	100,0%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 43, 615817
WDFY3	100,0%	100,0%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	98,1%	98,1%	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	100,0%	100,0%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	100,0%	100,0%	No OMIM Disease ID
WDR26	97,0%	94,3%	Skraban-Deardorff syndrome, 617616
WDR37	86,5%	86,5%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100,0%	100,0%	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45	100,0%	100,0%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	100,0%	100,0%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	100,0%	100,0%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	100,0%	100,0%	Galloway-Mowat syndrome 1, 251300
WDR81	100,0%	100,0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WFS1	100,0%	100,0%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400

			Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WWOX	100,0%	100,0%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XPA	100,0%	100,0%	Xeroderma pigmentosum, group A, 278700
XRCC4	100,0%	100,0%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100,0%	99,7%	Desbuquois dysplasia 2, 615777
YIF1B	90,1%	90,1%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100,0%	100,0%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	100,0%	100,0%	?Optic atrophy 11, 617302
YWHAE	100,0%	100,0%	No OMIM Disease ID
YWHAG	100,0%	100,0%	Developmental and epileptic encephalopathy 56, 617665
YY1	100,0%	100,0%	Gabriele-de Vries syndrome, 617557
ZBTB11	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100,0%	100,0%	Leukemia, acute promyelocytic, PL2F/RARA type,
ZBTB18	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 22, 612337
ZBTB20	100,0%	100,0%	Primrose syndrome, 259050
ZBTB24	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100,0%	100,0%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZDHHC9	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Raymond type, 300799
ZEB2	97,4%	97,4%	Mowat-Wilson syndrome, 235730
ZFHX4	100,0%	100,0%	No OMIM Disease ID
ZFYVE26	100,0%	100,0%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	100,0%	100,0%	?Craniosynostosis 6, 616602 Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100,0%	100,0%	Holoprosencephaly 5, 609637
ZMIZ1	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYM2	100,0%	100,0%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522
ZMYND11	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZNF142	100,0%	100,0%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	100,0%	100,0%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99,6%	99,6%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100,0%	100,0%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	100,0%	100,0%	SIMHA syndrome, 619557

ZNF41	100,0%	100,0%	No OMIM Disease ID
ZNF462	100,0%	100,0%	Weiss-Kruszka syndrome, 618619
ZNF526	100,0%	100,0%	No OMIM Disease ID
ZNF699	100,0%	100,0%	DEGCAGS syndrome, 619488
ZNF711	100,0%	100,0%	Intellectual developmental disorder, X-linked 97, 300803
ZSWIM6	97,6%	96,3%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 Acromelic frontonasal dysostosis, 603671

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.
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.
TWIST is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors