

INTELLECTUAL DISABILITY GENE PANEL DG 3.5.0 (1649 genes)

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<i>Gene</i>	<i>TWIST X2 covered >10x</i>	<i>TWIST X2 covered >20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
AAAS	100%	100%	<i>Achalasia-addisonianism-alacrimia syndrome, 231550</i>
AARS1	100%	100%	<i>Developmental and epileptic encephalopathy 29, 616339</i> <i>Charcot-Marie-Tooth disease, axonal, type 2N, 613287</i> <i>?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661</i> <i>Trichothiodystrophy 8, nonphotosensitive, 619691</i>
AASS	100%	100%	<i>Hyperlysinemia, 238700</i>
ABAT	100%	100%	<i>GABA-transaminase deficiency, 613163</i>
ABCA2	100%	100%	<i>Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808</i>
ABCC8	100%	100%	<i>Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857</i> <i>Diabetes mellitus, transient neonatal 2, 610374</i> <i>Diabetes mellitus, noninsulin-dependent, 125853</i> <i>Hypoglycemia of infancy, leucine-sensitive, 240800</i> <i>Hyperinsulinemic hypoglycemia, familial, 1, 256450</i>
ABCC9	100%	100%	<i>Cardiomyopathy, dilated, 10, 608569</i> <i>Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850</i> <i>?Atrial fibrillation, familial, 12, 614050</i> <i>Intellectual disability and myopathy syndrome, 619719</i>
ABCD1	100%	100%	<i>Adrenoleukodystrophy, 300100</i> <i>Adrenomyeloneuropathy, adult, 300100</i>
ABCD4	100%	100%	<i>Methylmalonic aciduria and homocystinuria, cblJ type, 614857</i>
ABHD16A	100%	100%	<i>Spastic paraplegia 86, autosomal recessive, 619735</i>
ABHD5	100%	100%	<i>Chanarin-Dorfman syndrome, 275630</i>
ACAD9	100%	100%	<i>Mitochondrial complex I deficiency, nuclear type 20, 611126</i>
ACADS	100%	100%	<i>Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470</i>
ACADSB	100%	100%	<i>2-methylbutyrylglycinuria, 610006</i>
ACAT1	100%	100%	<i>Alpha-methylacetoacetic aciduria, 203750</i>
ACER3	100%	100%	<i>?Leukodystrophy, progressive, early childhood-onset, 617762</i>
ACO2	100%	100%	<i>Optic atrophy 9, 616289</i> <i>Infantile cerebellar-retinal degeneration, 614559</i>
ACOX1	100%	100%	<i>Mitchell syndrome, 618960</i> <i>Peroxisomal acyl-CoA oxidase deficiency, 264470</i>
ACSF3	100%	100%	<i>Combined malonic and methylmalonic aciduria, 614265</i>
ACSL4	100%	100%	<i>Intellectual developmental disorder, X-linked 63, 300387</i>
ACTB	100%	100%	<i>Baraitser-Winter syndrome 1, 243310</i> <i>?Dystonia, juvenile-onset, 607371</i>

ACTG1	100%	100%	<i>Deafness, autosomal dominant 20/26, 604717</i> <i>Baraitser-Winter syndrome 2, 614583</i>
ACTL6A	100%	100%	<i>No OMIM disease ID</i>
ACTL6B	100%	100%	<i>Developmental and epileptic encephalopathy 76, 618468</i> <i>Intellectual developmental disorder with severe speech and ambulation defects, 618470</i>
ACVR1	100%	100%	<i>Fibrodysplasia ossificans progressiva, 135100</i>
ACY1	100%	100%	<i>Aminoacylase 1 deficiency, 609924</i>
ADAM22	100%	100%	<i>Developmental and epileptic encephalopathy 61, 617933</i>
ADAR	100%	100%	<i>Dyschromatosis symmetrica hereditaria, 127400</i> <i>Aicardi-Goutieres syndrome 6, 615010</i>
ADARB1	95%	95%	<i>Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862</i>
ADAT3	100%	100%	<i>Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286</i>
ADD1	100%	100%	<i>No OMIM disease ID</i>
ADD3	100%	100%	<i>Cerebral palsy, spastic quadriplegic, 3, 617008</i>
ADGRG1	100%	100%	<i>Polymicrogyria, bilateral frontoparietal, 606854</i> <i>Polymicrogyria, bilateral perisylvian, 615752</i>
ADK	91%	91%	<i>Hypermethioninemia due to adenosine kinase deficiency, 614300</i>
ADNP	100%	100%	<i>Helsmoortel-van der Aa syndrome, 615873</i>
ADPRS	100%	100%	<i>Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170</i>
ADSL	100%	100%	<i>Adenylosuccinase deficiency, 103050</i>
AFF2	100%	100%	<i>Intellectual developmental disorder, X-linked 109, 309548</i>
AFF3	100%	100%	<i>KINSSHIP syndrome, 619297</i>
AFF4	100%	100%	<i>CHOPS syndrome, 616368</i>
AFG3L2	100%	100%	<i>Spastic ataxia 5, autosomal recessive, 614487</i> <i>Optic atrophy 12, 618977</i> <i>Spinocerebellar ataxia 28, 610246</i>
AGA	100%	100%	<i>Aspartylglucosaminuria, 208400</i>
AGAP1	100%	100%	<i>No OMIM disease ID</i>
AGMO	100%	100%	<i>No OMIM disease ID</i>
AGO1	100%	100%	<i>No OMIM disease ID</i>
AGO2	100%	100%	<i>Lessel-Kreienkamp syndrome, 619149</i>
AGTPBP1	100%	100%	<i>Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276</i>
AHCY	100%	100%	<i>Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752</i>
AHDC1	100%	100%	<i>Xia-Gibbs syndrome, 615829</i>
AHI1	100%	100%	<i>Joubert syndrome 3, 608629</i>

AHSG	100%	100%	?Alopecia-intellectual disability syndrome 1, 203650
AIFM1	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
AIMP1	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100%	100%	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	100%	100%	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	94%	94%	Sjogren-Larsson syndrome, 270200
ALDH4A1	100%	100%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	100%	100%	Epilepsy, pyridoxine-dependent, 266100
ALG1	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96%	96%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100%	99%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100%	100%	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%	100%	Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96%	96%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type II, 608776
ALKBH8	100%	100%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100%	100%	Alstrom syndrome, 203800
ALX3	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451

AMER1	100%	100%	<i>Osteopathia striata with cranial sclerosis, 300373</i>
AMMECR1	100%	100%	<i>Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990</i>
AMPD2	100%	100%	? <i>Spastic paraplegia 63, 615686</i> <i>Pontocerebellar hypoplasia, type 9, 615809</i>
AMT	100%	100%	<i>Glycine encephalopathy, 605899</i>
ANK2	100%	100%	<i>Long QT syndrome 4, 600919</i> <i>Cardiac arrhythmia, ankyrin-B-related, 600919</i>
ANK3	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 37, 615493</i>
ANKH	100%	100%	<i>Chondrocalcinosis 2, 118600</i> <i>Cranio metaphyseal dysplasia, 123000</i>
ANKLE2	100%	100%	<i>Microcephaly 16, primary, autosomal recessive, 616681</i>
ANKRD11	100%	100%	<i>KBG syndrome, 148050</i>
ANKRD17	100%	100%	<i>Chopra-Amiel-Gordon syndrome, 619504</i>
ANKS1B	100%	100%	<i>No OMIM disease ID</i>
ANO10	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 10, 613728</i>
ANTXR1	100%	100%	<i>GAP0 syndrome, 230740</i>
AP1G1	100%	100%	<i>Usmani-Riazuddin syndrome, autosomal recessive, 619548</i> <i>Usmani-Riazuddin syndrome, autosomal dominant, 619467</i>
AP1S1	100%	100%	<i>MEDNIK syndrome, 609313</i>
AP1S2	100%	100%	<i>Pettigrew syndrome, 304340</i>
AP2M1	100%	100%	<i>Intellectual developmental disorder 60 with seizures, 618587</i>
AP2S1	100%	100%	<i>Hypocalciuric hypercalcemia, type III, 600740</i>
AP3B1	100%	100%	<i>Hermansky-Pudlak syndrome 2, 608233</i>
AP3B2	100%	100%	<i>Developmental and epileptic encephalopathy 48, 617276</i>
AP3D1	100%	100%	? <i>Hermansky-Pudlak syndrome 10, 617050</i>
AP4B1	100%	100%	<i>Spastic paraplegia 47, autosomal recessive, 614066</i>
AP4E1	100%	100%	<i>Stuttering, familial persistent, 1, 184450</i> <i>Spastic paraplegia 51, autosomal recessive, 613744</i>
AP4M1	100%	100%	<i>Spastic paraplegia 50, autosomal recessive, 612936</i>
AP4S1	87%	87%	<i>Spastic paraplegia 52, autosomal recessive, 614067</i>
APC2	100%	100%	<i>Cortical dysplasia, complex, with other brain malformations 10, 618677</i> <i>Intellectual developmental disorder, autosomal recessive 74, 617169</i>
APTX	100%	100%	<i>Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920</i>
ARCN1	100%	100%	<i>Short stature-micrognathia syndrome, 617164</i>
ARF1	100%	100%	<i>Periventricular nodular heterotopia 8, 618185</i>

ARF3	100%	100%	No OMIM disease ID
ARFGEF1	100%	100%	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964
ARFGEF2	100%	100%	Periventricular heterotopia with microcephaly, 608097
ARG1	93%	93%	Argininemia, 207800
ARHGAP31	100%	100%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100%	100%	No OMIM disease ID
ARHGEF6	100%	100%	No OMIM disease ID
ARHGEF9	97%	96%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100%	100%	Coffin-Siris syndrome 2, 614607
ARID1B	99%	98%	Coffin-Siris syndrome 1, 135900
ARID2	100%	100%	Coffin-Siris syndrome 6, 617808
ARL13B	100%	100%	Joubert syndrome 8, 612291
ARL6	100%	100%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC9	100%	100%	Joubert syndrome 30, 617622
ARPC4	100%	100%	No OMIM disease ID
ARSA	100%	100%	Metachromatic leukodystrophy, 250100
ARSL	100%	100%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100%	100%	Developmental and epileptic encephalopathy 38, 617020
ARX	99%	97%	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%	100%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASH1L	99%	99%	Intellectual developmental disorder, autosomal dominant 52, 617796
ASL	100%	100%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	Asparagine synthetase deficiency, 615574
ASPA	100%	100%	Canavan disease, 271900
ASPM	100%	100%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	100%	100%	Citrullinemia, 215700
ASXL1	100%	100%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039

ASXL2	100%	100%	<i>Shashi-Pena syndrome, 617190</i>
ASXL3	100%	100%	<i>Bainbridge-Ropers syndrome, 615485</i>
ATAD1	100%	100%	<i>Hyperekplexia 4, 618011</i>
ATAD3A	100%	100%	<i>Harel-Yoon syndrome, 617183</i> <i>Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810</i>
ATG7	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 31, 619422</i>
ATIC	100%	100%	<i>AICA-ribosiduria due to ATIC deficiency, 608688</i>
ATL1	100%	100%	<i>Spastic paraplegia 3A, autosomal dominant, 182600</i> <i>Neuropathy, hereditary sensory, type ID, 613708</i>
ATN1	100%	100%	<i>Dentatorubral-pallidoluysian atrophy, 125370</i> <i>Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494</i>
ATP13A2	100%	100%	<i>Spastic paraplegia 78, autosomal recessive, 617225</i> <i>Kufor-Rakeb syndrome, 606693</i>
ATP1A1	100%	100%	<i>Hypomagnesemia, seizures, and impaired intellectual development 2, 618314</i> <i>Charcot-Marie-Tooth disease, axonal, type 2DD, 618036</i>
ATP1A2	100%	100%	<i>Developmental and epileptic encephalopathy 98, 619605</i> <i>Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602</i> <i>Alternating hemiplegia of childhood 1, 104290</i> <i>Migraine, familial basilar, 602481</i> <i>Migraine, familial hemiplegic, 2, 602481</i>
ATP1A3	100%	100%	<i>Alternating hemiplegia of childhood 2, 614820</i> <i>Dystonia-12, 128235</i> <i>CAPOS syndrome, 601338</i> <i>Developmental and epileptic encephalopathy 99, 619606</i>
ATP2A2	100%	100%	<i>Acrokeratosis verruciformis, 101900</i> <i>Darier disease, 124200</i>
ATP2B1	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 66, 619910</i>
ATP6AP1	100%	100%	<i>Immunodeficiency 47, 300972</i>
ATP6AP2	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423</i> <i>?Parkinsonism with spasticity, X-linked, 300911</i> <i>Congenital disorder of glycosylation, type IIr, 301045</i>
ATP6VOA1	100%	100%	<i>Neurodevelopmental disorder with epilepsy and brain atrophy, 619971</i> <i>Developmental and epileptic encephalopathy 104, 619970</i>
ATP6VOA2	100%	100%	<i>Wrinkly skin syndrome, 278250</i> <i>Cutis laxa, autosomal recessive, type IIA, 219200</i>
ATP6VOC	100%	100%	<i>No OMIM disease ID</i>

ATP6V1A	100%	100%	<i>Cutis laxa, autosomal recessive, type IID, 617403</i> <i>Developmental and epileptic encephalopathy 93, 618012</i>
ATP6V1B2	100%	100%	<i>Zimmermann-Laband syndrome 2, 616455</i> <i>Deafness, congenital, with onychodystrophy, autosomal dominant, 124480</i>
ATP7A	100%	100%	<i>Occipital horn syndrome, 304150</i> <i>Spinal muscular atrophy, distal, X-linked 3, 300489</i> <i>Menkes disease, 309400</i>
ATP8A2	100%	100%	<i>?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268</i>
ATP9A	100%	100%	<i>No OMIM disease ID</i>
ATR	100%	100%	<i>Seckel syndrome 1, 210600</i> <i>?Cutaneous telangiectasia and cancer syndrome, familial, 614564</i>
ATRX	100%	100%	<i>Alpha-thalassemia/mental retardation syndrome, 301040</i> <i>Alpha-thalassemia myelodysplasia syndrome, somatic, 300448</i> <i>Intellectual disability-hypotonic facies syndrome, X-linked, 309580</i>
ATXN2L	100%	100%	<i>No OMIM disease ID</i>
AUH	100%	100%	<i>3-methylglutaconic aciduria, type I, 250950</i>
AUTS2	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 26, 615834</i>
AVPR2	100%	100%	<i>Diabetes insipidus, nephrogenic, 1, 304800</i> <i>Nephrogenic syndrome of inappropriate antidiuresis, 300539</i>
B3GALNT2	92%	92%	<i>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181</i>
B3GALT6	100%	98%	<i>Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349</i> <i>Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640</i> <i>Al-Gazali syndrome, 609465</i>
B3GLCT	100%	100%	<i>Peters-plus syndrome, 261540</i>
B4GALNT1	100%	100%	<i>Spastic paraplegia 26, autosomal recessive, 609195</i>
B4GALT1	100%	100%	<i>Congenital disorder of glycosylation, type IIc, 607091</i>
B4GALT7	100%	100%	<i>Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070</i>
B4GAT1	100%	100%	<i>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287</i>
B9D1	100%	100%	<i>?Meckel syndrome 9, 614209</i> <i>Joubert syndrome 27, 617120</i>
B9D2	100%	100%	<i>?Meckel syndrome 10, 614175</i> <i>Joubert syndrome 34, 614175</i>
BAP1	100%	100%	<i>Kury-Isidor syndrome, 619762</i> <i>Tumor predisposition syndrome 1, 614327</i>
BAZ2B	100%	100%	<i>No OMIM disease ID</i>
BBS1	100%	100%	<i>Bardet-Biedl syndrome 1, 209900</i>

BBS10	100%	100%	<i>Bardet-Biedl syndrome 10, 615987</i>
BBS12	100%	100%	<i>Bardet-Biedl syndrome 12, 615989</i>
BBS2	100%	100%	<i>Retinitis pigmentosa 74, 616562</i> <i>Bardet-Biedl syndrome 2, 615981</i>
BBS4	100%	100%	<i>Bardet-Biedl syndrome 4, 615982</i>
BBS5	100%	100%	<i>Bardet-Biedl syndrome 5, 615983</i>
BBS7	100%	100%	<i>Bardet-Biedl syndrome 7, 615984</i>
BBS9	96%	96%	<i>Bardet-Biedl syndrome 9, 615986</i>
BCAP31	99%	93%	<i>Deafness, dystonia, and cerebral hypomyelination, 300475</i>
BCAS3	100%	100%	<i>Hengel-Marooofian-Schols syndrome, 619641</i>
BCKDHA	100%	100%	<i>Maple syrup urine disease, type Ia, 248600</i>
BCKDHB	100%	100%	<i>Maple syrup urine disease, type Ib, 248600</i>
BCKDK	100%	100%	<i>Branched-chain keto acid dehydrogenase kinase deficiency, 614923</i>
BCL11A	100%	100%	<i>Dias-Logan syndrome, 617101</i>
BCL11B	100%	100%	<i>Immunodeficiency 49, severe combined, 617237</i> <i>Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092</i>
BCOR	100%	100%	<i>Microphthalmia, syndromic 2, 300166</i>
BCORL1	100%	100%	<i>Shukla-Vernon syndrome, 301029</i>
BCS1L	100%	100%	<i>GRACILE syndrome, 603358</i> <i>Mitochondrial complex III deficiency, nuclear type 1, 124000</i> <i>Bjornstad syndrome, 262000</i>
BICRA	100%	100%	<i>Coffin-Siris syndrome 12, 619325</i>
BLM	100%	100%	<i>Bloom syndrome, 210900</i>
BLOC1S1	100%	100%	<i>No OMIM disease ID</i>
KIAA1109	100%	100%	<i>Alkuraya-Kucinskas syndrome, 617822</i>
BOLA3	100%	100%	<i>Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299</i>
BPTF	100%	100%	<i>Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755</i>
BRAF	100%	100%	<i>Melanoma, malignant, somatic, 155600</i> <i>LEOPARD syndrome 3, 613707</i> <i>Cardiofaciocutaneous syndrome, 115150</i> <i>Adenocarcinoma of lung, somatic, 211980</i> <i>Noonan syndrome 7, 613706</i> <i>Colorectal cancer, somatic, 114500</i> <i>Nonsmall cell lung cancer, somatic, 211980</i>
BRAT1	100%	100%	<i>Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056</i> <i>Rigidity and multifocal seizure syndrome, lethal neonatal, 614498</i>

BRF1	100%	100%	<i>Cerebellofaciodental syndrome, 616202</i>
BRPF1	100%	100%	<i>Intellectual developmental disorder with dysmorphic facies and ptosis, 617333</i>
BRSK2	100%	100%	<i>No OMIM disease ID</i>
BRWD3	100%	100%	<i>Intellectual developmental disorder, X-linked 93, 300659</i>
BSCL2	100%	100%	<i>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</i>
BTD	94%	94%	<i>Biotinidase deficiency, 253260</i>
BUB1B	100%	100%	<i>Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300</i>
C12orf4	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 66, 618221</i>
C12orf57	100%	100%	<i>Temtamy syndrome, 218340</i>
C2CD3	96%	96%	<i>Orofaciodigital syndrome XIV, 615948</i>
CA2	100%	100%	<i>Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730</i>
CA5A	100%	100%	<i>Hyperammonemia due to carbonic anhydrase VA deficiency, 615751</i>
CA8	100%	100%	<i>Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227</i>
CACNA1A	100%	100%	<i>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</i>
CACNA1B	100%	100%	<i>Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497</i>
CACNA1C	100%	100%	<i>Timothy syndrome, 601005 Long QT syndrome 8, 618447 Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 Brugada syndrome 3, 611875</i>
CACNA1D	100%	100%	<i>Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896</i>
CACNA1E	100%	100%	<i>Developmental and epileptic encephalopathy 69, 618285</i>
CACNA1G	100%	100%	<i>Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087</i>
CACNA1I	100%	100%	<i>Neurodevelopmental disorder with speech impairment and with or without seizures, 620114</i>
CACNA2D1	100%	100%	<i>No OMIM disease ID</i>
CACNA2D2	100%	100%	<i>Cerebellar atrophy with seizures and variable developmental delay, 618501</i>
CAD	100%	100%	<i>Developmental and epileptic encephalopathy 50, 616457</i>

CAMK2A	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 53, 617798 ?Intellectual developmental disorder, autosomal recessive 63, 618095</i>
CAMK2B	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 54, 617799</i>
CAMK2G	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 59, 618522</i>
CAMK4	100%	100%	<i>No OMIM disease ID</i>
CAMTA1	100%	100%	<i>Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756</i>
CANT1	100%	100%	<i>Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719</i>
CAPN15	100%	100%	<i>Oculogastrointestinal neurodevelopmental syndrome, 619318</i>
CARS1	100%	100%	<i>Microcephaly, developmental delay, and brittle hair syndrome, 618891</i>
CARS2	100%	100%	<i>Combined oxidative phosphorylation deficiency 27, 616672</i>
CASK	100%	100%	<i>Intellectual developmental disorder, with or without nystagmus, 300422 Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422</i>
CBL	100%	100%	<i>Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785</i>
CBS	100%	100%	<i>Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200</i>
CC2D1A	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 3, 608443</i>
CC2D2A	98%	98%	<i>COACH syndrome 2, 619111 Retinitis pigmentosa 93, 619845 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285</i>
CCBE1	100%	100%	<i>Hennekam lymphangiectasia-lymphedema syndrome 1, 235510</i>
CCDC115	100%	100%	<i>Congenital disorder of glycosylation, type Ilo, 616828</i>
CCDC174	100%	100%	<i>Hypotonia, infantile, with psychomotor retardation, 616816</i>
CCDC186	100%	100%	<i>No OMIM disease ID</i>
CCDC22	100%	100%	<i>Ritscher-Schinzel syndrome 2, 300963</i>
CCDC32	100%	100%	<i>Cardiofacioneurodevelopmental syndrome, 619123</i>
CCDC47	100%	100%	<i>Trichohepatoneurodevelopmental syndrome, 618268</i>
CCDC88A	97%	97%	<i>?PEHO syndrome-like, 617507</i>
CCDC88C	100%	100%	<i>?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600</i>
CCND2	100%	100%	<i>Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938</i>
CCNK	99%	96%	<i>?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147</i>
CDC42	100%	100%	<i>Takenouchi-Kosaki syndrome, 616737</i>

CDC42BPB	100%	100%	<i>Chilton-Okur-Chung neurodevelopmental syndrome, 619841</i>
CDC6	100%	100%	<i>?Meier-Gorlin syndrome 5, 613805</i>
CDH11	100%	100%	<i>Teebi hypertelorism syndrome 2, 619736</i> <i>Elsahy-Waters syndrome, 211380</i>
CDH15	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 3, 612580</i>
CDH2	100%	100%	<i>Arrhythmogenic right ventricular dysplasia, familial, 14, 618920</i> <i>?Attention deficit-hyperactivity disorder 8, 619957</i> <i>Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929</i>
CDK10	100%	100%	<i>Al Kaissi syndrome, 617694</i>
CDK13	100%	100%	<i>Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360</i>
CDK19	100%	100%	<i>Developmental and epileptic encephalopathy 87, 618916</i>
CDK5RAP2	100%	100%	<i>Microcephaly 3, primary, autosomal recessive, 604804</i>
CDK8	100%	100%	<i>Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748</i>
CDKL5	96%	95%	<i>Developmental and epileptic encephalopathy 2, 300672</i>
CDKN1C	100%	100%	<i>IMAGE syndrome, 614732</i> <i>Beckwith-Wiedemann syndrome, 130650</i>
CDON	100%	100%	<i>Holoprosencephaly 11, 614226</i>
CELF2	100%	100%	<i>Developmental and epileptic encephalopathy 97, 619561</i>
CENPF	100%	100%	<i>Stromme syndrome, 243605</i>
CENPJ	100%	100%	<i>Microcephaly 6, primary, autosomal recessive, 608393</i> <i>?Seckel syndrome 4, 613676</i>
CEP104	100%	100%	<i>Joubert syndrome 25, 616781</i> <i>Intellectual developmental disorder, autosomal recessive 77, 619988</i>
CEP120	100%	100%	<i>Short-rib thoracic dysplasia 13 with or without polydactyly, 616300</i> <i>Joubert syndrome 31, 617761</i>
CEP135	100%	100%	<i>Microcephaly 8, primary, autosomal recessive, 614673</i>
CEP152	100%	100%	<i>Microcephaly 9, primary, autosomal recessive, 614852</i> <i>Seckel syndrome 5, 613823</i>
CEP290	100%	100%	<i>Leber congenital amaurosis 10, 611755</i> <i>Joubert syndrome 5, 610188</i> <i>Senior-Loken syndrome 6, 610189</i> <i>?Bardet-Biedl syndrome 14, 615991</i> <i>Meckel syndrome 4, 611134</i>
CEP41	100%	100%	<i>Joubert syndrome 15, 614464</i>
CEP55	100%	100%	<i>Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500</i>
CEP57	100%	100%	<i>Mosaic variegated aneuploidy syndrome 2, 614114</i>

CEP63	100%	100%	?Seckel syndrome 6, 614728
CEP83	100%	100%	Nephronophthisis 18, 615862
CEP85L	100%	100%	Lissencephaly 10, 618873
CEP89	100%	100%	No OMIM disease ID
CERT1	100%	100%	Intellectual developmental disorder, autosomal dominant 34, 616351
CHAMP1	100%	100%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579
CHD1	100%	100%	Pilarowski-Bjornsson syndrome, 617682
CHD2	100%	100%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100%	100%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100%	100%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	100%	100%	Intellectual developmental disorder with autism and macrocephaly, 615032
CHKA	100%	100%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023
CHKB	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CHRM1	100%	100%	No OMIM disease ID
CHRNA4	100%	100%	Epilepsy, nocturnal frontal lobe, 1, 600513
CIC	100%	100%	Intellectual developmental disorder, autosomal dominant 45, 617600
CIT	100%	100%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	100%	100%	Filippi syndrome, 272440
CLCN3	97%	97%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100%	100%	Raynaud-Claes syndrome, 300114
CLDN11	100%	100%	Leukodystrophy, hypomyelinating, 22, 619328
CLIC2	100%	100%	?Intellectual developmental disorder, X-linked syndromic 32, 300886
CLIP1	100%	100%	No OMIM disease ID
CLN3	93%	93%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83%	83%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100%	100%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143

CLP1	100%	100%	<i>Pontocerebellar hypoplasia, type 10, 615803</i>
CLPB	100%	100%	<i>Neutropenia, severe congenital, 9, autosomal dominant, 619813</i> <i>3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271</i> <i>3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835</i>
CLTC	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 56, 617854</i>
CNKS2	100%	99%	<i>Intellectual developmental disorder, X-linked syndromic, Houge type, 301008</i>
CNNM2	100%	100%	<i>Hypomagnesemia 6, renal, 613882</i> <i>Hypomagnesemia, seizures, and impaired intellectual development 1, 616418</i>
CNOT1	100%	100%	<i>Vissers-Bodmer syndrome, 619033</i> <i>Holoprosencephaly 12, with or without pancreatic agenesis, 618500</i>
CNOT2	100%	100%	<i>Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608</i>
CNOT3	100%	100%	<i>Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672</i>
CNPY3	100%	100%	<i>Developmental and epileptic encephalopathy 60, 617929</i>
CNTNAP1	100%	100%	<i>Lethal congenital contracture syndrome 7, 616286</i> <i>Hypomyelinating neuropathy, congenital, 3, 618186</i>
CNTNAP2	100%	100%	<i>Pitt-Hopkins like syndrome 1, 610042</i>
COA8	100%	100%	<i>Mitochondrial complex IV deficiency, nuclear type 17, 619061</i>
COASY	100%	100%	<i>Pontocerebellar hypoplasia, type 12, 618266</i> <i>Neurodegeneration with brain iron accumulation 6, 615643</i>
COG1	100%	100%	<i>Congenital disorder of glycosylation, type IIg, 611209</i>
COG4	100%	100%	<i>Congenital disorder of glycosylation, type IIj, 613489</i> <i>Saul-Wilson syndrome, 618150</i>
COG5	100%	100%	<i>Congenital disorder of glycosylation, type Ili, 613612</i>
COG6	100%	100%	<i>Shaheen syndrome, 615328</i> <i>Congenital disorder of glycosylation, type III, 614576</i>
COG7	100%	100%	<i>Congenital disorder of glycosylation, type ILe, 608779</i>
COG8	100%	100%	<i>Congenital disorder of glycosylation, type IIh, 611182</i>
COL4A1	100%	100%	<i>?Retinal arteries, tortuosity of, 180000</i> <i>Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773</i> <i>Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564</i> <i>Brain small vessel disease with or without ocular anomalies, 175780</i>
COL4A2	100%	100%	<i>Brain small vessel disease 2, 614483</i>
COLEC11	100%	100%	<i>3MC syndrome 2, 265050</i>
COPB1	100%	100%	<i>Baralle-Macken syndrome, 619255</i>
COPB2	100%	100%	<i>Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884</i> <i>?Microcephaly 19, primary, autosomal recessive, 617800</i>

COQ2	96%	96%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100%	100%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX15	100%	100%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100%	100%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX6B1	100%	100%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
CPE	100%	100%	BDV syndrome, 619326
CPLANE1	100%	100%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CPLX1	100%	100%	Developmental and epileptic encephalopathy 63, 617976
CPS1	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF3	100%	100%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876
CRADD	100%	100%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	100%	99%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREBBP	100%	100%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRLF1	100%	99%	Cold-induced sweating syndrome 1, 272430
CRPPA	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CSDE1	100%	100%	No OMIM disease ID
CSF1R	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSNK1G1	100%	100%	No OMIM disease ID
CSNK2A1	94%	94%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100%	100%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	100%	100%	Joubert syndrome 21, 615636
CSTB	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	100%	100%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100%	100%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTDP1	100%	100%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100%	99%	Cortical dysplasia, complex, with other brain malformations 9, 618174

CTNNB1	100%	100%	<i>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</i>
CTNND1	100%	100%	<i>Blepharocheilodontic syndrome 2, 617681</i>
CTNND2	100%	100%	<i>No OMIM disease ID</i>
CTSA	100%	100%	<i>Galactosialidosis, 256540</i>
CTSD	100%	100%	<i>Ceroid lipofuscinosis, neuronal, 10, 610127</i>
CTTNBP2	100%	100%	<i>No OMIM disease ID</i>
CTU2	100%	100%	<i>Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142</i>
CUL3	100%	100%	<i>Neurodevelopmental disorder with or without autism or seizures, 619239 Pseudohypoadosteronism, type IIE, 614496</i>
CUL4B	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354</i>
CUX1	100%	100%	<i>Global developmental delay with or without impaired intellectual development, 618330</i>
CUX2	100%	100%	<i>Developmental and epileptic encephalopathy 67, 618141</i>
CWC27	100%	100%	<i>Retinitis pigmentosa with or without skeletal anomalies, 250410</i>
CWF19L1	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 17, 616127</i>
CYB5R3	100%	100%	<i>Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800</i>
CYFIP2	100%	100%	<i>Developmental and epileptic encephalopathy 65, 618008</i>
CYP27A1	100%	100%	<i>Cerebrotendinous xanthomatosis, 213700</i>
CYP2U1	100%	100%	<i>Spastic paraplegia 56, autosomal recessive, 615030</i>
D2HGDH	100%	100%	<i>D-2-hydroxyglutaric aciduria, 600721</i>
DAG1	100%	100%	<i>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818</i>
DARS1	100%	100%	<i>Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281</i>
DARS2	100%	100%	<i>Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105</i>
DBT	100%	100%	<i>Maple syrup urine disease, type II, 248600</i>
DCAF17	100%	100%	<i>Woodhouse-Sakati syndrome, 241080</i>
DCC	100%	100%	<i>Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Esophageal carcinoma, somatic, 133239</i>

			<i>Colorectal cancer, somatic, 114500</i> <i>Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542</i>
<i>DCHS1</i>	<i>100%</i>	<i>100%</i>	<i>Mitral valve prolapse 2, 607829</i> <i>Van Maldergem syndrome 1, 601390</i>
<i>DCPS</i>	<i>100%</i>	<i>100%</i>	<i>Al-Raqad syndrome, 616459</i>
<i>DCX</i>	<i>99%</i>	<i>99%</i>	<i>Subcortical laminar heterotopia, X-linked, 300067</i> <i>Lissencephaly, X-linked, 300067</i>
<i>DDB1</i>	<i>100%</i>	<i>100%</i>	<i>White-Kernohan syndrome, 619426</i>
<i>DDC</i>	<i>100%</i>	<i>100%</i>	<i>Aromatic L-amino acid decarboxylase deficiency, 608643</i>
<i>DDHD2</i>	<i>100%</i>	<i>100%</i>	<i>Spastic paraplegia 54, autosomal recessive, 615033</i>
<i>DDX11</i>	<i>100%</i>	<i>100%</i>	<i>Warsaw breakage syndrome, 613398</i>
<i>DDX23</i>	<i>100%</i>	<i>100%</i>	<i>No OMIM disease ID</i>
<i>DDX3X</i>	<i>99%</i>	<i>98%</i>	<i>Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958</i>
<i>DDX59</i>	<i>100%</i>	<i>100%</i>	<i>Orofaciodigital syndrome V, 174300</i>
<i>DDX6</i>	<i>100%</i>	<i>100%</i>	<i>Intellectual developmental disorder with impaired language and dysmorphic facies, 618653</i>
<i>DEAF1</i>	<i>100%</i>	<i>100%</i>	<i>Vulto-van Silfout-de Vries syndrome, 615828</i> <i>Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171</i>
<i>DEGS1</i>	<i>100%</i>	<i>100%</i>	<i>Leukodystrophy, hypomyelinating, 18, 618404</i>
<i>DENND5A</i>	<i>100%</i>	<i>100%</i>	<i>Developmental and epileptic encephalopathy 49, 617281</i>
<i>DEPDC5</i>	<i>100%</i>	<i>100%</i>	<i>Epilepsy, familial focal, with variable foci 1, 604364</i>
<i>DHCR24</i>	<i>100%</i>	<i>100%</i>	<i>Desmosterolosis, 602398</i>
<i>DHCR7</i>	<i>100%</i>	<i>100%</i>	<i>Smith-Lemli-Opitz syndrome, 270400</i>
<i>DHDDS</i>	<i>94%</i>	<i>94%</i>	<i>Developmental delay and seizures with or without movement abnormalities, 617836</i> <i>?Congenital disorder of glycosylation, type 1bb, 613861</i> <i>Retinitis pigmentosa 59, 613861</i>
<i>DHFR</i>	<i>100%</i>	<i>100%</i>	<i>Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839</i>
<i>DHPS</i>	<i>97%</i>	<i>93%</i>	<i>Neurodevelopmental disorder with seizures and speech and walking impairment, 618480</i>
<i>DHTKD1</i>	<i>100%</i>	<i>100%</i>	<i>?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025</i> <i>Alpha-aminoacidic and alpha-ketoacidic aciduria, 204750</i>
<i>DHX16</i>	<i>100%</i>	<i>100%</i>	<i>Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733</i>
<i>DHX30</i>	<i>100%</i>	<i>100%</i>	<i>Neurodevelopmental disorder with severe motor impairment and absent language, 617804</i>
<i>DHX37</i>	<i>100%</i>	<i>100%</i>	<i>Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731</i> <i>46, XY sex reversal 11, 273250</i>
<i>DIAPH1</i>	<i>100%</i>	<i>100%</i>	<i>Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900</i> <i>Seizures, cortical blindness, microcephaly syndrome, 616632</i>
<i>DIP2B</i>	<i>100%</i>	<i>100%</i>	<i>Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630</i>

<i>DIS3L2</i>	100%	100%	<i>Perlman syndrome, 267000</i>
<i>DKC1</i>	100%	100%	<i>Dyskeratosis congenita, X-linked, 305000</i>
<i>DLAT</i>	100%	100%	<i>Pyruvate dehydrogenase E2 deficiency, 245348</i>
<i>DLD</i>	100%	100%	<i>Dihydrolipoamide dehydrogenase deficiency, 246900</i>
<i>DLG3</i>	100%	100%	<i>Intellectual developmental disorder, X-linked 90, 300850</i>
<i>DLG4</i>	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 62, 618793</i>
<i>DLL1</i>	100%	100%	<i>Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709</i>
<i>DMD</i>	100%	99%	<i>Becker muscular dystrophy, 300376</i> <i>Cardiomyopathy, dilated, 3B, 302045</i> <i>Duchenne muscular dystrophy, 310200</i>
<i>DMPK</i>	100%	100%	<i>Myotonic dystrophy 1, 160900</i>
<i>DMXL2</i>	100%	100%	<i>Developmental and epileptic encephalopathy 81, 618663</i> <i>?Deafness, autosomal dominant 71, 617605</i> <i>?Polyendocrine-polyneuropathy syndrome, 616113</i>
<i>DNAJC12</i>	100%	100%	<i>Hyperphenylalaninemia, mild, non-BH4-deficient, 617384</i>
<i>DNAJC19</i>	100%	100%	<i>3-methylglutaconic aciduria, type V, 610198</i>
<i>DNM1</i>	100%	100%	<i>Developmental and epileptic encephalopathy 31, 616346</i>
<i>DNM1L</i>	100%	100%	<i>Optic atrophy 5, 610708</i> <i>Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388</i>
<i>DNMT3A</i>	100%	100%	<i>Tatton-Brown-Rahman syndrome, 615879</i> <i>Acute myeloid leukemia, somatic, 601626</i> <i>Heyn-Sproul-Jackson syndrome, 618724</i>
<i>DNMT3B</i>	100%	100%	<i>Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860</i> <i>Facioscapulohumeral muscular dystrophy 4, digenic, 619478</i>
<i>DOCK3</i>	100%	100%	<i>Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292</i>
<i>DOCK6</i>	100%	100%	<i>Adams-Oliver syndrome 2, 614219</i>
<i>DOCK7</i>	100%	100%	<i>Developmental and epileptic encephalopathy 23, 615859</i>
<i>DOHH</i>	100%	100%	<i>Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066</i>
<i>DOLK</i>	100%	100%	<i>Congenital disorder of glycosylation, type Im, 610768</i>
<i>DONSON</i>	100%	100%	<i>Microcephaly, short stature, and limb abnormalities, 617604</i> <i>Microcephaly-micromelia syndrome, 251230</i>
<i>DPAGT1</i>	100%	100%	<i>Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750</i> <i>Congenital disorder of glycosylation, type Ij, 608093</i>
<i>DPF2</i>	100%	100%	<i>Coffin-Siris syndrome 7, 618027</i>
<i>DPH1</i>	100%	100%	<i>Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901</i>
<i>DPH5</i>	100%	100%	<i>Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070</i>

DPM1	99%	97%	<i>Congenital disorder of glycosylation, type Ie, 608799</i>
DPM2	100%	100%	<i>Congenital disorder of glycosylation, type Iu, 615042</i>
DPP6	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 33, 616311</i>
DPYD	100%	100%	<i>Dihydropyrimidine dehydrogenase deficiency, 274270</i> <i>5-fluorouracil toxicity, 274270</i>
DPYS	100%	100%	<i>Dihydropyrimidinuria, 222748</i>
DPYSL5	100%	100%	<i>Ritscher-Schinzel syndrome 4, 619435</i>
DTYMK	100%	100%	<i>Neurodegeneration, childhood-onset, with progressive microcephaly, 619847</i>
DYM	100%	100%	<i>Smith-McCort dysplasia, 607326</i> <i>Dyggve-Melchior-Clausen disease, 223800</i>
DYNC1H1	100%	100%	<i>Charcot-Marie-Tooth disease, axonal, type 2O, 614228</i> <i>Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600</i> <i>Intellectual developmental disorder, autosomal dominant 13, 614563</i>
DYNC1I2	100%	100%	<i>Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492</i>
DYRK1A	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 7, 614104</i>
EARS2	100%	100%	<i>Combined oxidative phosphorylation deficiency 12, 614924</i>
EBF3	100%	100%	<i>Hypotonia, ataxia, and delayed development syndrome, 617330</i>
EBP	100%	100%	<i>MEND syndrome, 300960</i> <i>Chondrodysplasia punctata, X-linked dominant, 302960</i>
ECHS1	100%	100%	<i>Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277</i>
EDC3	100%	100%	<i>?Intellectual developmental disorder, autosomal recessive 50, 616460</i>
EDEM3	100%	100%	<i>Congenital disorder of glycosylation, type IIv, 619493</i>
EED	100%	100%	<i>Cohen-Gibson syndrome, 617561</i>
EEF1A2	100%	100%	<i>Developmental and epileptic encephalopathy 33, 616409</i> <i>Intellectual developmental disorder, autosomal dominant 38, 616393</i>
EFNB2	100%	100%	<i>No OMIM disease ID</i>
EFTUD2	100%	100%	<i>Mandibulofacial dysostosis, Guion-Almeida type, 610536</i>
EHMT1	100%	100%	<i>Kleefstra syndrome 1, 610253</i>
EIF2AK1	100%	100%	<i>?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878</i>
EIF2AK2	100%	100%	<i>Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877</i> <i>Dystonia 33, 619687</i>
EIF2AK3	100%	100%	<i>Wolcott-Rallison syndrome, 226980</i>
EIF2B4	100%	100%	<i>Ovarioleukodystrophy, 603896</i> <i>Leukoencephalopathy with vanishing white matter, 603896</i>
EIF2B5	100%	100%	<i>Ovarioleukodystrophy, 603896</i> <i>Leukoencephalopathy with vanishing white matter, 603896</i>

EIF2S3	100%	100%	MEHMO syndrome, 300148
EIF3F	100%	100%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A3	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF5A	100%	100%	Faundes-Banka syndrome, 619376
ELAC2	100%	100%	Combined oxidative phosphorylation deficiency 17, 615440
ELOVL4	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELP2	100%	100%	Intellectual developmental disorder, autosomal recessive 58, 617270
EMC1	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100%	100%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EML1	100%	100%	Band heterotopia, 600348
EMX2	100%	100%	Schizencephaly, 269160
ENTPD1	100%	100%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	100%	100%	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPG5	100%	100%	Vici syndrome, 242840
EPHA7	100%	100%	No OMIM disease ID
ERCC1	100%	100%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100%	100%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC5	100%	100%	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100%	100%	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%	100%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400

ERLIN2	100%	100%	<i>Spastic paraplegia 18, autosomal recessive, 611225</i>
ESCO2	100%	100%	<i>Juberg-Hayward syndrome, 216100</i> <i>Roberts-SC phocomelia syndrome, 268300</i>
ETFB	100%	100%	<i>Glutaric acidemia IIB, 231680</i>
ETHE1	100%	100%	<i>Ethylmalonic encephalopathy, 602473</i>
EXOC2	100%	100%	<i>Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306</i>
EXOC7	100%	100%	<i>Neurodevelopmental disorder with seizures and brain atrophy, 619072</i>
EXOSC2	100%	100%	<i>Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763</i>
EXOSC3	100%	100%	<i>Pontocerebellar hypoplasia, type 1B, 614678</i>
EXOSC8	100%	100%	<i>Pontocerebellar hypoplasia, type 1C, 616081</i>
EXOSC9	100%	100%	<i>Pontocerebellar hypoplasia, type 1D, 618065</i>
EXTL3	100%	100%	<i>Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425</i>
EZH2	100%	100%	<i>Weaver syndrome, 277590</i>
FA2H	100%	100%	<i>Spastic paraplegia 35, autosomal recessive, 612319</i>
FAM149B1	100%	100%	<i>Joubert syndrome 36, 618763</i>
FAM20C	100%	100%	<i>Raine syndrome, 259775</i>
FAM50A	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261</i>
FAR1	100%	100%	<i>Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154</i> <i>Cataracts, spastic paraparesis, and speech delay, 619338</i>
FARS2	100%	100%	<i>Combined oxidative phosphorylation deficiency 14, 614946</i> <i>Spastic paraplegia 77, autosomal recessive, 617046</i>
FARSB	100%	100%	<i>Rajab interstitial lung disease with brain calcifications 1, 613658</i>
FAT4	100%	100%	<i>Van Maldergem syndrome 2, 615546</i> <i>Hennekam lymphangiectasia-lymphedema syndrome 2, 616006</i>
FBRSL1	100%	99%	<i>No OMIM disease ID</i>
FBXL3	100%	100%	<i>Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220</i>
FBXL4	100%	100%	<i>Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471</i>
FBXO11	100%	100%	<i>Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089</i>
FBXO28	100%	100%	<i>Developmental and epileptic encephalopathy 100, 619777</i>
FBXO31	100%	100%	<i>?Intellectual developmental disorder, autosomal recessive 45, 615979</i>
FBXW11	100%	100%	<i>Neurodevelopmental, jaw, eye, and digital syndrome, 618914</i>
FBXW7	100%	98%	<i>Developmental delay, hypotonia, and impaired language, 620012</i>
FDFT1	100%	100%	<i>Squalene synthase deficiency, 618156</i>
FGD1	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic 16, 305400</i> <i>Aarskog-Scott syndrome, 305400</i>

FGF12	100%	100%	<i>Developmental and epileptic encephalopathy 47, 617166</i>
FGF13	100%	100%	<i>Developmental and epileptic encephalopathy 90, 301058</i>
FGF14	100%	100%	<i>Spinocerebellar ataxia 27, 193003</i>
FGFR1	100%	100%	<i>Pfeiffer syndrome, 101600</i> <i>Hypogonadotropic hypogonadism 2 with or without anosmia, 147950</i> <i>Jackson-Weiss syndrome, 123150</i> <i>Hartsfield syndrome, 615465</i> <i>Trigonocephaly 1, 190440</i> <i>Osteoglophonic dysplasia, 166250</i> <i>Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001</i>
FGFR2	100%	100%	<i>Bent bone dysplasia syndrome, 614592</i> <i>LADD syndrome, 149730</i> <i>Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410</i> <i>Jackson-Weiss syndrome, 123150</i> <i>Gastric cancer, somatic, 613659</i> <i>Craniofacial-skeletal-dermatologic dysplasia, 101600</i> <i>Apert syndrome, 101200</i> <i>Pfeiffer syndrome, 101600</i> <i>?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579</i> <i>Beare-Stevenson cutis gyrata syndrome, 123790</i> <i>Crouzon syndrome, 123500</i> <i>Saethre-Chotzen syndrome, 101400</i> <i>Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,</i>
FGFR3	100%	100%	<i>Muenke syndrome, 602849</i> <i>SADDAN, 616482</i> <i>Hypochondroplasia, 146000</i> <i>LADD syndrome, 149730</i> <i>Thanatophoric dysplasia, type II, 187601</i> <i>Nevus, epidermal, somatic, 162900</i> <i>CATSHL syndrome, 610474</i> <i>Thanatophoric dysplasia, type I, 187600</i> <i>Spermatocytic seminoma, somatic, 273300</i> <i>Bladder cancer, somatic, 109800</i> <i>Achondroplasia, 100800</i> <i>Cervical cancer, somatic, 603956</i> <i>Colorectal cancer, somatic, 114500</i> <i>Crouzon syndrome with acanthosis nigricans, 612247</i>

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

FRAS1	100%	100%	Fraser syndrome 1, 219000
FRMD4A	97%	97%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMPD4	100%	100%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100%	100%	Developmental and epileptic encephalopathy 37, 616981
FTCD	100%	100%	Glutamate formiminotransferase deficiency, 229100
FTO	95%	95%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	100%	100%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100%	100%	Fucosidosis, 230000
FUT8	100%	100%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FZR1	100%	100%	No OMIM disease ID
GABBR2	100%	100%	Developmental and epileptic encephalopathy 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903
GABRA1	100%	100%	Developmental and epileptic encephalopathy 19, 615744
GABRA2	100%	100%	Developmental and epileptic encephalopathy 78, 618557
GABRA3	100%	100%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRA5	100%	100%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100%	100%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100%	100%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	100%	100%	Developmental and epileptic encephalopathy 43, 617113
GABRD	100%	100%	No OMIM disease ID
GABRG2	93%	93%	Developmental and epileptic encephalopathy 74, 618396 Febrile seizures, familial, 8, 607681 Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100%	100%	Developmental and epileptic encephalopathy 89, 619124
GALC	100%	100%	Krabbe disease, 245200
GALE	100%	100%	Galactose epimerase deficiency, 230350
GALNT2	100%	100%	Congenital disorder of glycosylation, type IIc, 618885
GALT	100%	100%	Galactosemia, 230400
GAMT	100%	100%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	100%	100%	GAND syndrome, 615074
GATM	100%	100%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GCH1	100%	100%	Dystonia, DOPA-responsive, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	100%	100%	?Glycine encephalopathy, 605899

<i>GDI1</i>	100%	100%	<i>Intellectual developmental disorder, X-linked 41, 300849</i>
<i>GEMIN5</i>	100%	100%	<i>Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333</i>
<i>GFAP</i>	100%	100%	<i>Alexander disease, 203450</i>
<i>GFER</i>	100%	100%	<i>Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076</i>
<i>GFM1</i>	100%	100%	<i>Combined oxidative phosphorylation deficiency 1, 609060</i>
<i>GFM2</i>	100%	100%	<i>Combined oxidative phosphorylation deficiency 39, 618397</i>
<i>GIGYF1</i>	100%	100%	<i>No OMIM disease ID</i>
<i>GJA1</i>	100%	100%	<i>Erythrokeratoderma variabilis et progressiva 3, 617525</i> <i>Craniometaphyseal dysplasia, autosomal recessive, 218400</i> <i>Oculodentodigital dysplasia, 164200</i> <i>Hypoplastic left heart syndrome 1, 241550</i> <i>Palmoplantar keratoderma with congenital alopecia, 104100</i> <i>Syndactyly, type III, 186100</i> <i>Oculodentodigital dysplasia, autosomal recessive, 257850</i> <i>Atrioventricular septal defect 3, 600309</i>
<i>GJB1</i>	100%	100%	<i>Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800</i>
<i>GJC2</i>	100%	99%	<i>Lymphatic malformation 3, 613480</i> <i>?Spastic paraplegia 44, autosomal recessive, 613206</i> <i>Leukodystrophy, hypomyelinating, 2, 608804</i>
<i>GK</i>	100%	100%	<i>Glycerol kinase deficiency, 307030</i>
<i>GLB1</i>	100%	100%	<i>GM1-gangliosidosis, type I, 230500</i> <i>GM1-gangliosidosis, type III, 230650</i> <i>Mucopolysaccharidosis type IVB (Morquio), 253010</i> <i>GM1-gangliosidosis, type II, 230600</i>
<i>GLDC</i>	100%	100%	<i>Glycine encephalopathy, 605899</i>
<i>GLI2</i>	100%	100%	<i>Culler-Jones syndrome, 615849</i> <i>Holoprosencephaly 9, 610829</i>
<i>GLI3</i>	100%	100%	<i>Greig cephalopolysyndactyly syndrome, 175700</i> <i>Polydactyly, postaxial, types A1 and B, 174200</i> <i>Pallister-Hall syndrome, 146510</i> <i>Polydactyly, preaxial, type IV, 174700</i>
<i>GLIS3</i>	100%	100%	<i>Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199</i>
<i>GLRA2</i>	100%	98%	<i>Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076</i>
<i>GLS</i>	100%	100%	<i>Global developmental delay, progressive ataxia, and elevated glutamine, 618412</i> <i>?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339</i> <i>Developmental and epileptic encephalopathy 71, 618328</i>

GLUD1	100%	100%	<i>Hyperinsulinism-hyperammonemia syndrome, 606762</i>
GLUL	100%	100%	<i>Glutamine deficiency, congenital, 610015</i>
GLYCK	100%	100%	<i>D-glyceric aciduria, 220120</i>
GM2A	100%	100%	<i>GM2-gangliosidosis, AB variant, 272750</i>
GMNN	100%	100%	<i>Meier-Gorlin syndrome 6, 616835</i>
GMPPA	100%	100%	<i>Alacrima, achalasia, and impaired intellectual development syndrome, 615510</i>
GMPPB	100%	100%	<i>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352</i> <i>Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351</i> <i>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350</i>
GNAI1	100%	100%	<i>Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854</i>
GNAO1	100%	100%	<i>Developmental and epileptic encephalopathy 17, 615473</i> <i>Neurodevelopmental disorder with involuntary movements, 617493</i>
GNAS	100%	100%	<i>ACTH-independent macronodular adrenal hyperplasia, 219080</i> <i>Pituitary adenoma 3, multiple types, somatic, 617686</i> <i>Pseudohypoparathyroidism 1c, 612462</i> <i>Pseudohypoparathyroidism 1a, 103580</i> <i>Osseous heteroplasia, progressive, 166350</i> <i>Pseudohypoparathyroidism 1b, 603233</i> <i>McCune-Albright syndrome, somatic, mosaic, 174800</i> <i>Pseudopseudohypoparathyroidism, 612463</i>
GNB1	100%	100%	<i>Myelodysplastic syndrome, somatic, 614286</i> <i>Leukemia, acute lymphoblastic, somatic, 613065</i> <i>Intellectual developmental disorder, autosomal dominant 42, 616973</i>
GNB2	100%	100%	<i>Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503</i> <i>?Sick sinus syndrome 4, 619464</i>
GNB5	100%	100%	<i>Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182</i> <i>Intellectual developmental disorder with cardiac arrhythmia, 617173</i>
GNPAT	100%	100%	<i>Rhizomelic chondrodysplasia punctata, type 2, 222765</i>
GNPTAB	100%	100%	<i>Mucopolidosis III alpha/beta, 252600</i> <i>Mucopolidosis II alpha/beta, 252500</i>
GNPTG	100%	100%	<i>Mucopolidosis III gamma, 252605</i>
GNS	100%	100%	<i>Mucopolysaccharidosis type IIID, 252940</i>
GOLGA2	100%	100%	<i>No OMIM disease ID</i>
GOT2	100%	100%	<i>Developmental and epileptic encephalopathy 82, 618721</i>
GPAA1	100%	100%	<i>Glycosylphosphatidylinositol biosynthesis defect 15, 617810</i>

GPC3	100%	99%	<i>Wilms tumor, somatic, 194070</i> <i>Simpson-Golabi-Behmel syndrome, type 1, 312870</i>
GPC4	100%	100%	<i>Keipert syndrome, 301026</i>
GPHN	100%	100%	<i>Molybdenum cofactor deficiency C, 615501</i>
GPSM2	100%	100%	<i>Chudley-McCullough syndrome, 604213</i>
GPT2	100%	100%	<i>Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281</i>
GRIA2	100%	100%	<i>Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917</i>
GRIA3	100%	99%	<i>Intellectual developmental disorder, X-linked syndromic, Wu type, 300699</i>
GRIA4	100%	100%	<i>Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864</i>
GRID2	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 18, 616204</i>
GRIK2	96%	96%	<i>Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580</i> <i>Intellectual developmental disorder, autosomal recessive 6, 611092</i>
GRIN1	100%	100%	<i>Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820</i> <i>Developmental and epileptic encephalopathy 101, 619814</i> <i>Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254</i>
GRIN2A	100%	99%	<i>Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570</i>
GRIN2B	100%	100%	<i>Developmental and epileptic encephalopathy 27, 616139</i> <i>Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970</i>
GRIN2D	100%	99%	<i>Developmental and epileptic encephalopathy 46, 617162</i>
GRIP1	100%	100%	<i>Fraser syndrome 3, 617667</i>
GRM1	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 13, 614831</i> <i>Spinocerebellar ataxia 44, 617691</i>
GRM7	100%	100%	<i>Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922</i>
GRN	100%	100%	<i>Aphasia, primary progressive, 607485</i> <i>Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485</i> <i>Ceroid lipofuscinosis, neuronal, 11, 614706</i>
GSE1	100%	100%	<i>No OMIM disease ID</i>
GSS	100%	100%	<i>Hemolytic anemia due to glutathione synthetase deficiency, 231900</i> <i>Glutathione synthetase deficiency, 266130</i>
GTF2E2	100%	100%	<i>Trichothiodystrophy 6, nonphotosensitive, 616943</i>
GTF2H5	70%	70%	<i>Trichothiodystrophy 3, photosensitive, 616395</i>
GTPBP2	100%	100%	<i>Jaberi-Elahi syndrome, 617988</i>
GTPBP3	100%	100%	<i>Combined oxidative phosphorylation deficiency 23, 616198</i>
GUSB	100%	100%	<i>Mucopolysaccharidosis VII, 253220</i>
H1-4	100%	100%	<i>Rahman syndrome, 617537</i>
H3-3B	100%	100%	<i>Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721</i>

H4C3	100%	100%	<i>Tessadori-van Haaften neurodevelopmental syndrome 1, 619758</i>
HAAO	100%	100%	<i>Vertebral, cardiac, renal, and limb defects syndrome 1, 617660</i>
HACE1	100%	100%	<i>Spastic paraplegia and psychomotor retardation with or without seizures, 616756</i>
HADH	100%	100%	<i>Hyperinsulinemic hypoglycemia, familial, 4, 609975</i> <i>3-hydroxyacyl-CoA dehydrogenase deficiency, 231530</i>
HADHA	100%	100%	<i>HELLP syndrome, maternal, of pregnancy, 609016</i> <i>Mitochondrial trifunctional protein deficiency, 609015</i> <i>LCHAD deficiency, 609016</i> <i>Fatty liver, acute, of pregnancy, 609016</i>
HADHB	100%	100%	<i>Trifunctional protein deficiency, 609015</i>
HAX1	100%	100%	<i>Neutropenia, severe congenital 3, autosomal recessive, 610738</i>
HCCS	100%	100%	<i>Linear skin defects with multiple congenital anomalies 1, 309801</i>
HCFC1	100%	100%	<i>Methylmalonic aciduria and homocysteinemia, cblX type, 309541</i>
HCN1	100%	100%	<i>Developmental and epileptic encephalopathy 24, 615871</i> <i>Generalized epilepsy with febrile seizures plus, type 10, 618482</i>
HDAC4	100%	100%	<i>Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797</i>
HDAC6	100%	100%	<i>?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863</i>
HDAC8	98%	97%	<i>Cornelia de Lange syndrome 5, 300882</i>
HEATR3	100%	100%	<i>Diamond-Blackfan anemia 21, 620072</i>
HEATR5B	100%	100%	<i>No OMIM disease ID</i>
HECW2	100%	100%	<i>Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268</i>
HEPACAM	100%	100%	<i>Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925</i> <i>Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926</i>
HERC1	100%	100%	<i>Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011</i>
HERC2	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 38, 615516</i>
HESX1	100%	100%	<i>Pituitary hormone deficiency, combined, 5, 182230</i> <i>Septooptic dysplasia, 182230</i> <i>Growth hormone deficiency with pituitary anomalies, 182230</i>
HEXA	100%	100%	<i>GM2-gangliosidosis, several forms, 272800</i> <i>Tay-Sachs disease, 272800</i>
HEXB	100%	100%	<i>Sandhoff disease, infantile, juvenile, and adult forms, 268800</i>
HGSNAT	92%	92%	<i>Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930</i> <i>Retinitis pigmentosa 73, 616544</i>
HIBCH	100%	100%	<i>3-hydroxyisobutryl-CoA hydrolase deficiency, 250620</i>
HID1	100%	100%	<i>Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983</i>
HIVEP2	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 43, 616977</i>

HK1	100%	100%	<i>Retinitis pigmentosa 79, 617460</i> <i>Neuropathy, hereditary motor and sensory, Russe type, 605285</i> <i>Neurodevelopmental disorder with visual defects and brain anomalies, 618547</i> <i>Hemolytic anemia due to hexokinase deficiency, 235700</i>
HLCS	100%	100%	<i>Holocarboxylase synthetase deficiency, 253270</i>
HMGB1	100%	100%	<i>No OMIM disease ID</i>
HMGCL	100%	100%	<i>HMG-CoA lyase deficiency, 246450</i>
HNMT	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 51, 616739</i>
HNRNPD	100%	100%	<i>No OMIM disease ID</i>
HNRNPH1	100%	100%	<i>Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083</i>
HNRNPH2	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Bain type, 300986</i>
HNRNPK	100%	100%	<i>Au-Kline syndrome, 616580</i>
HNRNPU	100%	100%	<i>Developmental and epileptic encephalopathy 54, 617391</i>
HOXA1	100%	100%	<i>Bosley-Salih-Alorainy syndrome, 601536</i> <i>Athabaskan brainstem dysgenesis syndrome, 601536</i>
HPD	100%	100%	<i>Hawkinsinuria, 140350</i> <i>Tyrosinemia, type III, 276710</i>
HPDL	100%	100%	<i>Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026</i> <i>Spastic paraplegia 83, autosomal recessive, 619027</i>
HPRT1	100%	100%	<i>Hyperuricemia, HRPT-related, 300323</i> <i>Lesch-Nyhan syndrome, 300322</i>
HRAS	100%	100%	<i>Bladder cancer, somatic, 109800</i> <i>Thyroid carcinoma, follicular, somatic, 188470</i> <i>Congenital myopathy with excess of muscle spindles, 218040</i> <i>Nevus sebaceous or woolly hair nevus, somatic, 162900</i> <i>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200</i> <i>Spitz nevus or nevus spilus, somatic, 137550</i> <i>Costello syndrome, 218040</i>
HS2ST1	100%	100%	<i>Neurofacioskeletal syndrome with or without renal agenesis, 619194</i>
HSD17B10	100%	100%	<i>HSD10 mitochondrial disease, 300438</i>
HSD17B4	97%	97%	<i>D-bifunctional protein deficiency, 261515</i> <i>Perrault syndrome 1, 233400</i>
HSPA9	100%	100%	<i>Even-plus syndrome, 616854</i> <i>Anemia, sideroblastic, 4, 182170</i>
HSPD1	100%	100%	<i>Spastic paraplegia 13, autosomal dominant, 605280</i> <i>Leukodystrophy, hypomyelinating, 4, 612233</i>

HTRA2	100%	100%	3-methylglutaconic aciduria, type VIII, 617248
HUWE1	100%	100%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590
FAM126A	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
HYLS1	100%	100%	Hydrolethalus syndrome, 236680
IARS1	100%	100%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100%	100%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
IDS	100%	100%	Mucopolysaccharidosis II, 309900
IDUA	100%	100%	Mucopolysaccharidosis I _s , 607016 Mucopolysaccharidosis I _{h/s} , 607015 Mucopolysaccharidosis I _h , 607014
IER3IP1	100%	100%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100%	100%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFT172	100%	100%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	Bardet-Biedl syndrome 19, 615996
IFT74	100%	100%	Bardet-Biedl syndrome 22, 617119 Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582
IFT81	95%	95%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	100%	100%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100%	100%	Insulin-like growth factor I, resistance to, 270450
IKBKG	100%	98%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636 Autoinflammatory disease, systemic, X-linked, 301081
IL1RAPL1	100%	100%	Intellectual developmental disorder, X-linked 21, 300143
IMPA1	100%	100%	Intellectual developmental disorder, autosomal recessive 59, 617323
INPP5E	100%	100%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156

INPP5K	100%	100%	<i>Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404</i>
INTS1	100%	100%	<i>Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571</i>
IPO8	100%	100%	<i>VISS syndrome, 619472</i>
IQSEC1	100%	100%	<i>Intellectual developmental disorder with short stature and behavioral abnormalities, 618687</i>
IQSEC2	100%	98%	<i>Intellectual developmental disorder, X-linked 1, 309530</i>
IREB2	100%	100%	<i>Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451</i>
IRF2BPL	100%	100%	<i>Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088</i>
IRX5	100%	100%	<i>Hamamy syndrome, 611174</i>
ISCA2	100%	100%	<i>Multiple mitochondrial dysfunctions syndrome 4, 616370</i>
ITGA7	100%	100%	<i>Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204</i>
ITPA	100%	100%	<i>Developmental and epileptic encephalopathy 35, 616647</i>
ITPR1	100%	100%	<i>Gillespie syndrome, 206700</i> <i>Spinocerebellar ataxia 29, congenital nonprogressive, 117360</i> <i>Spinocerebellar ataxia 15, 606658</i>
IVD	100%	100%	<i>Isovaleric acidemia, 243500</i>
JAG1	100%	100%	<i>?Deafness, congenital heart defects, and posterior embryotoxon, 617992</i> <i>Charcot-Marie-Tooth disease, axonal, type 2HH, 619574</i> <i>Alagille syndrome 1, 118450</i> <i>Tetralogy of Fallot, 187500</i>
JAG2	100%	100%	<i>Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566</i>
JAM3	100%	100%	<i>Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730</i>
JARID2	100%	100%	<i>Developmental delay with variable intellectual disability and dysmorphic facies, 620098</i>
JMJD1C	100%	100%	<i>No OMIM disease ID</i>
KANK1	100%	100%	<i>Cerebral palsy, spastic quadriplegic, 2, 612900</i>
KANSL1	100%	100%	<i>Koolen-De Vries syndrome, 610443</i>
KAT5	100%	100%	<i>Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103</i>
KAT6A	100%	100%	<i>Arboleda-Tham syndrome, 616268</i>
KAT6B	100%	100%	<i>SBBYSS syndrome, 603736</i> <i>Genitopatellar syndrome, 606170</i>
KAT8	100%	100%	<i>Li-Ghorgani-Weisz-Hubshman syndrome, 618974</i>
KATNB1	100%	100%	<i>Lissencephaly 6, with microcephaly, 616212</i>
KCNA2	100%	100%	<i>Developmental and epileptic encephalopathy 32, 616366</i>
KCNA4	100%	100%	<i>Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284</i>
KCNB1	100%	100%	<i>Developmental and epileptic encephalopathy 26, 616056</i>
KCNC1	100%	100%	<i>Epilepsy, progressive myoclonic 7, 616187</i>

KCNC3	100%	98%	<i>Spinocerebellar ataxia 13, 605259</i>
KCNH1	99%	99%	<i>Zimmermann-Laband syndrome 1, 135500</i> <i>Temple-Baraitser syndrome, 611816</i>
KCNJ10	100%	100%	<i>Enlarged vestibular aqueduct, digenic, 600791</i> <i>SESAME syndrome, 612780</i>
KCNJ11	100%	100%	<i>Diabetes, permanent neonatal 2, with or without neurologic features, 618856</i> <i>Maturity-onset diabetes of the young, type 13, 616329</i> <i>Diabetes mellitus, transient neonatal 3, 610582</i> <i>Hyperinsulinemic hypoglycemia, familial, 2, 601820</i>
KCNJ6	100%	100%	<i>Keppen-Lubinsky syndrome, 614098</i>
KCNK4	100%	100%	<i>Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381</i>
KCNK9	100%	100%	<i>Birk-Barel syndrome, 612292</i>
KCNMA1	100%	100%	<i>Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446</i> <i>Cerebellar atrophy, developmental delay, and seizures, 617643</i> <i>Liang-Wang syndrome, 618729</i>
KCNN2	100%	100%	<i>?Dystonia 34, myoclonic, 619724</i> <i>Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725</i>
KCNN3	100%	100%	<i>Zimmermann-Laband syndrome 3, 618658</i>
KCNQ2	100%	100%	<i>Developmental and epileptic encephalopathy 7, 613720</i> <i>Seizures, benign neonatal, 1, 121200</i> <i>Myokymia, 121200</i>
KCNQ3	100%	100%	<i>Seizures, benign neonatal, 2, 121201</i>
KCNQ5	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 46, 617601</i>
KCNT1	100%	100%	<i>Developmental and epileptic encephalopathy 14, 614959</i> <i>Epilepsy nocturnal frontal lobe, 5, 615005</i>
KCNT2	100%	99%	<i>Developmental and epileptic encephalopathy 57, 617771</i>
KCTD3	100%	100%	<i>No OMIM disease ID</i>
KCTD7	100%	100%	<i>Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726</i>
KDM1A	100%	100%	<i>Cleft palate, psychomotor retardation, and distinctive facial features, 616728</i>
KDM3B	100%	100%	<i>Diets-Jongmans syndrome, 618846</i>
KDM4B	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 65, 619320</i>
KDM5B	98%	96%	<i>Intellectual developmental disorder, autosomal recessive 65, 618109</i>
KDM5C	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534</i>
KDM6A	100%	100%	<i>Kabuki syndrome 2, 300867</i>
KDM6B	100%	100%	<i>Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505</i>

KIAA0586	96%	96%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIDINS220	100%	100%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 Ventriculomegaly and arthrogryposis, 619501
KIF11	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	100%	100%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	100%	100%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF21B	100%	100%	No OMIM disease ID
KIF2A	100%	100%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100%	100%	Retinitis pigmentosa 89, 618955
KIF4A	100%	100%	?Intellectual developmental disorder, X-linked 100, 300923
KIF5A	100%	100%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	99%	99%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100%	100%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydroletharus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96%	96%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL3	100%	100%	No OMIM disease ID
KLF7	100%	100%	No OMIM disease ID
KLHL15	100%	100%	Intellectual developmental disorder, X-linked 103, 300982
KMT2A	100%	100%	Wiedemann-Steiner syndrome, 605130
KMT2B	100%	100%	Intellectual developmental disorder, autosomal dominant 68, 619934 Dystonia 28, childhood-onset, 617284
KMT2C	100%	100%	Kleefstra syndrome 2, 617768
KMT2D	100%	100%	Kabuki syndrome 1, 147920
KMT2E	100%	100%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100%	100%	Intellectual developmental disorder, autosomal dominant 51, 617788
KNL1	99%	99%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100%	100%	Intellectual developmental disorder, autosomal recessive 41, 615637

KRAS	100%	100%	<i>Gastric cancer, somatic, 613659</i> <i>Oculoectodermal syndrome, somatic, 600268</i> <i>Breast cancer, somatic, 114480</i> <i>Noonan syndrome 3, 609942</i> <i>RAS-associated autoimmune leukoproliferative disorder, 614470</i> <i>Arteriovenous malformation of the brain, somatic, 108010</i> <i>Lung cancer, somatic, 211980</i> <i>Pancreatic carcinoma, somatic, 260350</i> <i>Leukemia, acute myeloid, somatic, 601626</i> <i>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200</i> <i>Cardiofaciocutaneous syndrome 2, 615278</i> <i>Bladder cancer, somatic, 109800</i>
L1CAM	100%	100%	<i>MASA syndrome, 303350</i> <i>Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000</i> <i>Corpus callosum, partial agenesis of, 304100</i> <i>CRASH syndrome, 303350</i> <i>Hydrocephalus with Hirschsprung disease, 307000</i> <i>Hydrocephalus due to aqueductal stenosis, 307000</i>
L2HGDH	100%	100%	<i>L-2-hydroxyglutaric aciduria, 236792</i>
LAMA1	100%	100%	<i>Poretti-Boltshauser syndrome, 615960</i>
LAMA2	100%	100%	<i>Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138</i> <i>Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855</i>
LAMB1	100%	100%	<i>Lissencephaly 5, 615191</i>
LAMB2	100%	100%	<i>Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199</i> <i>Pierson syndrome, 609049</i>
LAMC3	100%	100%	<i>Cortical malformations, occipital, 614115</i>
LAMP2	100%	100%	<i>Danon disease, 300257</i>
LARGE1	100%	100%	<i>Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840</i> <i>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154</i>
LARP7	100%	100%	<i>Alazami syndrome, 615071</i>
LARS1	100%	100%	<i>?Infantile liver failure syndrome 1, 615438</i>
LAS1L	100%	100%	<i>Wilson-Turner syndrome, 309585</i>
LIAS	100%	100%	<i>Hyperglycinemia, lactic acidosis, and seizures, 614462</i>
LIG4	100%	100%	<i>LIG4 syndrome, 606593</i>
LINGO1	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 64, 618103</i>
LINS1	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 27, 614340</i>

LMAN2L	100%	100%	?Intellectual developmental disorder, autosomal dominant 69, 617863 ?Intellectual developmental disorder, autosomal recessive 52, 616887
LMBRD2	100%	100%	Developmental delay with variable neurologic and brain abnormalities, 619694
LMNB1	100%	100%	Leukodystrophy, adult-onset, autosomal dominant, 169500 Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	100%	100%	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LONP1	100%	100%	CODAS syndrome, 600373
LRP2	100%	100%	Donnai-Barrow syndrome, 222448
LRPPRC	100%	100%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LSS	100%	100%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-intellectual disability syndrome 4, 618840
LYRM7	100%	100%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	100%	100%	Chediak-Higashi syndrome, 214500
LZTFL1	100%	100%	Bardet-Biedl syndrome 17, 615994
LZTR1	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAB21L1	100%	100%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100%	100%	Lissencephaly 9 with complex brainstem malformation, 618325
MADD	100%	100%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 DEEAH syndrome, 619004
MAF	94%	90%	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088
MAG	100%	100%	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	100%	100%	Schaaf-Yang syndrome, 615547
MAN1B1	100%	100%	Rafiq syndrome, 614202
MAN2B1	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MAN2C1	100%	100%	Congenital disorder of deglycosylation 2, 619775
MANBA	100%	100%	Mannosidosis, beta, 248510
MAOA	99%	99%	Brunner syndrome, 300615
MAP1B	100%	100%	?Deafness, autosomal dominant 83, 619808 Periventricular nodular heterotopia 9, 618918
MAP2K1	100%	100%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950

MAP2K2	100%	100%	<i>Cardiofaciocutaneous syndrome 4, 615280</i>
MAPK1	100%	100%	<i>Noonan syndrome 13, 619087</i>
MAPK8IP3	100%	100%	<i>Neurodevelopmental disorder with or without variable brain abnormalities, 618443</i>
MAPKAPK5	100%	100%	<i>Neurocardiofaciodigital syndrome, 619869</i>
MAPRE2	100%	100%	<i>Symmetric circumferential skin creases, congenital, 2, 616734</i>
MASP1	100%	100%	<i>3MC syndrome 1, 257920</i>
MAST1	100%	100%	<i>Mega-corpora-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273</i>
MAT1A	100%	100%	<i>Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850</i> <i>Methionine adenosyltransferase deficiency, autosomal recessive, 250850</i>
MBD5	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 1, 156200</i>
MBOAT7	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 57, 617188</i>
MBTPS2	100%	100%	<i>Keratosis follicularis spinulosa decalvans, X-linked, 308800</i> <i>Osteogenesis imperfecta, type XIX, 301014</i> <i>IFAP syndrome with or without BRESHECK syndrome, 308205</i> <i>?Olmsted syndrome, X-linked, 300918</i>
MCCC1	100%	100%	<i>3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200</i>
MCCC2	100%	100%	<i>3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210</i>
MCOLN1	100%	100%	<i>Mucopolidosis IV, 252650</i>
MCPH1	100%	100%	<i>Microcephaly 1, primary, autosomal recessive, 251200</i>
MDH2	100%	100%	<i>Developmental and epileptic encephalopathy 51, 617339</i>
MECP2	100%	100%	<i>Rett syndrome, atypical, 312750</i> <i>Encephalopathy, neonatal severe, 300673</i> <i>Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260</i> <i>Intellectual developmental disorder, X-linked syndromic 13, 300055</i> <i>Rett syndrome, 312750</i> <i>Rett syndrome, preserved speech variant, 312750</i>
MECR	100%	100%	<i>Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282</i>
MED12	100%	100%	<i>Lujan-Fryns syndrome, 309520</i> <i>Ohdo syndrome, X-linked, 300895</i> <i>Hardikar syndrome, 301068</i> <i>Opitz-Kaveggia syndrome, 305450</i>
MED12L	100%	100%	<i>Nizon-Isidor syndrome, 618872</i>
MED13	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 61, 618009</i>
MED13L	100%	100%	<i>Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789</i>
MED17	100%	100%	<i>Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668</i>
MED23	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249</i>

MED25	100%	100%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MED27	100%	100%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEF2C	100%	100%	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEGF8	100%	100%	Carpenter syndrome 2, 614976
MEIS2	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
METTL23	100%	100%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100%	100%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	100%	100%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MGP	100%	100%	Keutel syndrome, 245150
MIA3	100%	100%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MICU1	100%	100%	Myopathy with extrapyramidal signs, 615673
MID1	100%	99%	Opitz GBBB syndrome, 300000
MID2	100%	100%	?Intellectual developmental disorder, X-linked 101, 300928
MINPP1	100%	100%	Pontocerebellar hypoplasia, type 16, 619527
MKKS	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	100%	100%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MLC1	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MLYCD	100%	100%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, cbIA type, 251100
MMAB	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, cbIB type, 251110
MMACHC	100%	100%	Methylmalonic aciduria and homocystinuria, cbIC type, 277400
MMADHC	89%	89%	Methylmalonic aciduria, cbID type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cbID type, 277410 Homocystinuria, cbID type, variant 1, 277410
MMGT1	100%	100%	No OMIM disease ID
MMUT	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100%	100%	CEBALID syndrome, 618774 Meningioma, 607174

MOCS1	100%	100%	<i>Molybdenum cofactor deficiency A, 252150</i>
MOCS2	100%	100%	<i>Molybdenum cofactor deficiency B, 252160</i>
MOGS	100%	100%	<i>Congenital disorder of glycosylation, type IIb, 606056</i>
MORC2	100%	100%	<i>Charcot-Marie-Tooth disease, axonal, type 2Z, 616688</i> <i>Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090</i>
MPDU1	100%	100%	<i>Congenital disorder of glycosylation, type If, 609180</i>
MPDZ	100%	99%	<i>Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219</i>
MPLKIP	100%	100%	<i>Trichothiodystrophy 4, nonphotosensitive, 234050</i>
MPV17	100%	100%	<i>Charcot-Marie-Tooth disease, axonal, type 2EE, 618400</i> <i>Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810</i>
MRAS	100%	100%	<i>Noonan syndrome 11, 618499</i>
MRPS22	100%	100%	<i>Ovarian dysgenesis 7, 618117</i> <i>Combined oxidative phosphorylation deficiency 5, 611719</i>
MRPS34	100%	100%	<i>Combined oxidative phosphorylation deficiency 32, 617664</i>
MSL2	100%	100%	<i>No OMIM disease ID</i>
MSL3	100%	100%	<i>Basilicata-Akhtar syndrome, 301032</i>
MSMO1	100%	100%	<i>Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834</i>
MTFMT	100%	100%	<i>Combined oxidative phosphorylation deficiency 15, 614947</i> <i>Mitochondrial complex I deficiency, nuclear type 27, 618248</i>
MTHFR	100%	100%	<i>Homocystinuria due to MTHFR deficiency, 236250</i>
MTHFS	100%	100%	<i>Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367</i>
MTO1	94%	91%	<i>Combined oxidative phosphorylation deficiency 10, 614702</i>
MTOR	100%	100%	<i>Focal cortical dysplasia, type II, somatic, 607341</i> <i>Smith-Kingsmore syndrome, 616638</i>
MTR	100%	100%	<i>Homocystinuria-megaloblastic anemia, cblG complementation type, 250940</i>
C12orf65	100%	100%	<i>Spastic paraplegia 55, autosomal recessive, 615035</i> <i>Combined oxidative phosphorylation deficiency 7, 613559</i>
MTRR	100%	100%	<i>Homocystinuria-megaloblastic anemia, cbl E type, 236270</i>
MTSS2	100%	100%	<i>Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086</i>
MVK	90%	90%	<i>Hyper-IgD syndrome, 260920</i> <i>Porokeratosis 3, multiple types, 175900</i> <i>Mevalonic aciduria, 610377</i>
MYCN	100%	100%	<i>Feingold syndrome 1, 164280</i>
MYH9	100%	100%	<i>Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100</i> <i>Deafness, autosomal dominant 17, 603622</i>

MYO5A	100%	100%	Griscelli syndrome, type 1, 214450
MYO9A	100%	100%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYT1L	100%	100%	Intellectual developmental disorder, autosomal dominant 39, 616521
NAA10	100%	100%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	97%	97%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	100%	100%	Intellectual developmental disorder, autosomal recessive 73, 619717
NACC1	100%	100%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAGA	100%	100%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	100%	100%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	100%	100%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NAPB	100%	100%	Developmental and epileptic encephalopathy 107, 620033
NARS1	100%	100%	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%	100%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAXE	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	100%	99%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBN	100%	100%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCAPG2	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NCDN	100%	100%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCKAP1	100%	100%	No OMIM disease ID
NDE1	100%	100%	Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDP	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	100%	100%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100%	99%	Mitochondrial complex I deficiency, nuclear type 14, 618236

NDUFA12	100%	100%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	100%	100%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA8	100%	100%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFAF3	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	100%	100%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	97%	91%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100%	100%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	100%	100%	Periventricular nodular heterotopia 7, 617201
NEMF	100%	100%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEU1	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROD2	100%	100%	Developmental and epileptic encephalopathy 72, 618374
NEXMIF	100%	100%	Intellectual developmental disorder, X-linked 98, 300912
NF1	100%	100%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFE2L2	100%	100%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	100%	100%	Brain malformations with or without urinary tract defects, 613735
NFIB	100%	100%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100%	100%	Marshall-Smith syndrome, 602535 Malan syndrome, 614753
NFU1	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100%	100%	Congenital disorder of deglycosylation 1, 615273
NHLRC2	100%	100%	FINCA syndrome, 618278

NHS	100%	100%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIPBL	100%	100%	Cornelia de Lange syndrome 1, 122470
NKAP	100%	100%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NKX2-1	100%	100%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN2	100%	100%	No OMIM disease ID
NLGN3	100%	100%	No OMIM disease ID
NLGN4X	100%	100%	Intellectual developmental disorder, X-linked, 300495
NONO	100%	100%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOVA2	100%	100%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPC1	100%	100%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NR2F1	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	100%	100%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911
NRAS	100%	100%	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 mosaicism, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
NRCAM	100%	100%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833
NRROS	100%	100%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	100%	100%	Sotos syndrome, 117550
NSD2	100%	100%	Rauch-Steindl syndrome, 619695
NSDHL	100%	100%	CK syndrome, 300831 CHILD syndrome, 308050
NSF	100%	100%	Developmental and epileptic encephalopathy 96, 619340
NSRP1	91%	91%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001

NSUN2	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 5, 611091</i>
NT5C2	100%	100%	<i>Spastic paraplegia 45, autosomal recessive, 613162</i>
NTNG2	100%	100%	<i>Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718</i>
NTRK1	100%	100%	<i>Insensitivity to pain, congenital, with anhidrosis, 256800</i>
NTRK2	100%	100%	<i>Developmental and epileptic encephalopathy 58, 617830</i> <i>Obesity, hyperphagia, and developmental delay, 613886</i>
NUBPL	100%	100%	<i>Mitochondrial complex I deficiency, nuclear type 21, 618242</i>
NUDT2	100%	100%	<i>Intellectual developmental disorder with or without peripheral neuropathy, 619844</i>
NUP107	100%	100%	? <i>Ovarian dysgenesis 6, 618078</i> <i>Galloway-Mowat syndrome 7, 618348</i> <i>Nephrotic syndrome, type 11, 616730</i>
NUP188	100%	100%	<i>Sandestig-Stefanova syndrome, 618804</i>
NUP214	100%	100%	<i>Leukemia, T-cell acute lymphoblastic, somatic, 613065</i> <i>Leukemia, acute myeloid, somatic, 601626</i>
NUP62	100%	100%	<i>Striatonigral degeneration, infantile, 271930</i>
NUP85	100%	100%	<i>Nephrotic syndrome, type 17, 618176</i>
NUS1	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831</i> <i>?Congenital disorder of glycosylation, type 1aa, 617082</i>
OAT	100%	100%	<i>Gyrate atrophy of choroid and retina with or without ornithinemia, 258870</i>
OCLN	100%	100%	<i>Pseudo-TORCH syndrome 1, 251290</i>
OCRL	100%	100%	<i>Dent disease 2, 300555</i> <i>Lowe syndrome, 309000</i>
ODC1	100%	100%	<i>Bachmann-Bupp syndrome, 619075</i>
OFD1	100%	100%	<i>Simpson-Golabi-Behmel syndrome, type 2, 300209</i> <i>?Retinitis pigmentosa 23, 300424</i> <i>Orofaciodigital syndrome I, 311200</i> <i>Joubert syndrome 10, 300804</i>
OGDHL	100%	100%	<i>Yoon-Bellen neurodevelopmental syndrome, 619701</i>
OGT	100%	100%	<i>Intellectual developmental disorder, X-linked 106, 300997</i>
OPA3	100%	100%	<i>3-methylglutaconic aciduria, type III, 258501</i> <i>Optic atrophy 3 with cataract, 165300</i>
OPHN1	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486</i>
ORC1	100%	100%	<i>Meier-Gorlin syndrome 1, 224690</i>
OSGEP	100%	100%	<i>Galloway-Mowat syndrome 3, 617729</i>
OTC	100%	100%	<i>Ornithine transcarbamylase deficiency, 311250</i>

OTUD5	100%	100%	<i>Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056</i>
OTUD6B	100%	100%	<i>Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452</i>
OTUD7A	100%	98%	<i>No OMIM disease ID</i>
OTX2	100%	100%	<i>Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125</i> <i>Pituitary hormone deficiency, combined, 6, 613986</i> <i>Microphthalmia, syndromic 5, 610125</i>
OXR1	100%	100%	<i>Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000</i>
P4HTM	100%	100%	<i>Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493</i>
PACS1	100%	100%	<i>Schuurs-Hoeijmakers syndrome, 615009</i>
PACS2	100%	100%	<i>Developmental and epileptic encephalopathy 66, 618067</i>
PAFAH1B1	100%	100%	<i>Subcortical laminar heterotopia, 607432</i> <i>Lissencephaly 1, 607432</i>
PAH	100%	100%	<i>Phenylketonuria, 261600</i>
PAK1	100%	100%	<i>Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158</i>
PAK3	100%	99%	<i>Intellectual developmental disorder, X-linked 30, 300558</i>
MPP5	100%	100%	<i>No OMIM disease ID</i>
PAM16	85%	85%	<i>Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320</i>
PAN2	100%	100%	<i>No OMIM disease ID</i>
PANK2	100%	100%	<i>HARP syndrome, 607236</i> <i>Neurodegeneration with brain iron accumulation 1, 234200</i>
PANX1	100%	100%	<i>Oocyte maturation defect 7, 618550</i>
PARN	97%	96%	<i>Dyskeratosis congenita, autosomal recessive 6, 616353</i> <i>Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371</i>
PARP6	100%	100%	<i>No OMIM disease ID</i>
PAX1	100%	100%	<i>Otofaciocervical syndrome 2, 615560</i>
PAX6	100%	100%	<i>Optic nerve hypoplasia, 165550</i> <i>Cataract with late-onset corneal dystrophy, 106210</i> <i>?Coloboma, ocular, 120200</i> <i>?Coloboma of optic nerve, 120430</i> <i>Aniridia, 106210</i> <i>Anterior segment dysgenesis 5, multiple subtypes, 604229</i> <i>?Morning glory disc anomaly, 120430</i> <i>Foveal hypoplasia 1, 136520</i> <i>Keratitis, 148190</i>

PAX7	100%	100%	<i>Rhabdomyosarcoma 2, alveolar, 268220</i> <i>Myopathy, congenital, progressive, with scoliosis, 618578</i>
PAX8	100%	100%	<i>Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700</i>
PBX1	100%	100%	<i>Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641</i>
PC	100%	100%	<i>Pyruvate carboxylase deficiency, 266150</i>
PCCA	100%	100%	<i>Propionicacidemia, 606054</i>
PCCB	100%	98%	<i>Propionicacidemia, 606054</i>
PCDH12	100%	100%	<i>Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280</i>
PCDH19	100%	100%	<i>Developmental and epileptic encephalopathy 9, 300088</i>
PCDHGC4	100%	100%	<i>Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880</i>
PCGF2	100%	100%	<i>Turnpenny-Fry syndrome, 618371</i>
PCLO	100%	100%	<i>?Pontocerebellar hypoplasia, type 3, 608027</i>
PCNT	100%	100%	<i>Microcephalic osteodysplastic primordial dwarfism, type II, 210720</i>
PCYT2	100%	100%	<i>Spastic paraplegia 82, autosomal recessive, 618770</i>
PDE2A	100%	100%	<i>Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150</i>
PDE4D	100%	100%	<i>Acrodysostosis 2, with or without hormone resistance, 614613</i>
PDGFRB	100%	100%	<i>Premature aging syndrome, Penttinen type, 601812</i> <i>Kosaki overgrowth syndrome, 616592</i> <i>Myofibromatosis, infantile, 1, 228550</i> <i>Basal ganglia calcification, idiopathic, 4, 615007</i>
PDHA1	100%	98%	<i>Pyruvate dehydrogenase E1-alpha deficiency, 312170</i>
PDHB	100%	100%	<i>Pyruvate dehydrogenase E1-beta deficiency, 614111</i>
PDHX	100%	100%	<i>Lacticacidemia due to PDX1 deficiency, 245349</i>
PDP1	100%	100%	<i>Pyruvate dehydrogenase phosphatase deficiency, 608782</i>
PDSS1	100%	100%	<i>Coenzyme Q10 deficiency, primary, 2, 614651</i>
PDSS2	100%	100%	<i>Coenzyme Q10 deficiency, primary, 3, 614652</i>
PDZD8	100%	100%	<i>Intellectual developmental disorder with autism and dysmorphic facies, 620021</i>
PEPD	100%	100%	<i>Prolidase deficiency, 170100</i>
PET100	100%	100%	<i>Mitochondrial complex IV deficiency, nuclear type 12, 619055</i>
PEX1	100%	100%	<i>Heimler syndrome 1, 234580</i> <i>Peroxisome biogenesis disorder 1B (NALD/IRD), 601539</i> <i>Peroxisome biogenesis disorder 1A (Zellweger), 214100</i>
PEX10	100%	100%	<i>Peroxisome biogenesis disorder 6A (Zellweger), 614870</i> <i>Peroxisome biogenesis disorder 6B, 614871</i>

PEX11B	100%	100%	<i>Peroxisome biogenesis disorder 14B, 614920</i>
PEX12	100%	100%	<i>Peroxisome biogenesis disorder 3B, 266510</i> <i>Peroxisome biogenesis disorder 3A (Zellweger), 614859</i>
PEX13	100%	100%	<i>Peroxisome biogenesis disorder 11A (Zellweger), 614883</i> <i>Peroxisome biogenesis disorder 11B, 614885</i>
PEX16	100%	100%	<i>Peroxisome biogenesis disorder 8B, 614877</i> <i>Peroxisome biogenesis disorder 8A (Zellweger), 614876</i>
PEX19	100%	100%	<i>Peroxisome biogenesis disorder 12A (Zellweger), 614886</i>
PEX2	100%	100%	<i>Peroxisome biogenesis disorder 5A (Zellweger), 614866</i> <i>Peroxisome biogenesis disorder 5B, 614867</i>
PEX26	100%	100%	<i>Peroxisome biogenesis disorder 7B, 614873</i> <i>Peroxisome biogenesis disorder 7A (Zellweger), 614872</i>
PEX3	100%	100%	<i>Peroxisome biogenesis disorder 10A (Zellweger), 614882</i> <i>?Peroxisome biogenesis disorder 10B, 617370</i>
PEX5	100%	100%	<i>Peroxisome biogenesis disorder 2B, 202370</i> <i>Peroxisome biogenesis disorder 2A (Zellweger), 214110</i> <i>Rhizomelic chondrodysplasia punctata, type 5, 616716</i>
PEX6	100%	100%	<i>Peroxisome biogenesis disorder 4B, 614863</i> <i>Peroxisome biogenesis disorder 4A (Zellweger), 614862</i> <i>Heimler syndrome 2, 616617</i>
PEX7	91%	91%	<i>Rhizomelic chondrodysplasia punctata, type 1, 215100</i> <i>Peroxisome biogenesis disorder 9B, 614879</i>
PGAP1	100%	100%	<i>Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802</i>
PGAP2	100%	100%	<i>Hyperphosphatasia with impaired intellectual development syndrome 3, 614207</i>
PGAP3	100%	100%	<i>Hyperphosphatasia with impaired intellectual development syndrome 4, 615716</i>
PGK1	100%	100%	<i>Phosphoglycerate kinase 1 deficiency, 300653</i>
PGM2L1	100%	100%	<i>No OMIM disease ID</i>
PGM3	100%	100%	<i>Immunodeficiency 23, 615816</i>
PHACTR1	100%	100%	<i>Developmental and epileptic encephalopathy 70, 618298</i>
PHF21A	100%	100%	<i>Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725</i>
PHF6	100%	100%	<i>Borjeson-Forssman-Lehmann syndrome, 301900</i>
PHF8	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263</i>
PHGDH	100%	100%	<i>Neu-Laxova syndrome 1, 256520</i> <i>Phosphoglycerate dehydrogenase deficiency, 601815</i>
PHIP	100%	100%	<i>Chung-Jansen syndrome, 617991</i>

PI4KA	100%	100%	<i>Spastic paraplegia 84, autosomal recessive, 619621</i> <i>Gastrointestinal defects and immunodeficiency syndrome 2, 619708</i> <i>Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogyriposis, 616531</i>
PIBF1	100%	100%	<i>Joubert syndrome 33, 617767</i>
PIDD1	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827</i>
PIGA	100%	100%	<i>Paroxysmal nocturnal hemoglobinuria, somatic, 300818</i> <i>Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868</i> <i>Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072</i>
PIGB	100%	100%	<i>Developmental and epileptic encephalopathy 80, 618580</i>
PIGC	100%	100%	<i>Glycosylphosphatidylinositol biosynthesis defect 16, 617816</i>
PIGF	100%	100%	<i>Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356</i>
PIGG	100%	100%	<i>Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917</i>
PIGH	81%	75%	<i>Glycosylphosphatidylinositol biosynthesis defect 17, 618010</i>
PIGK	100%	100%	<i>Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879</i>
PIGL	100%	100%	<i>CHIME syndrome, 280000</i>
PIGN	100%	100%	<i>Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080</i>
PIGO	100%	100%	<i>Hyperphosphatasia with impaired intellectual development syndrome 2, 614749</i>
PIGP	100%	100%	<i>Developmental and epileptic encephalopathy 55, 617599</i>
PIGQ	100%	100%	<i>Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548</i>
PIGS	100%	100%	<i>Developmental and epileptic encephalopathy 95, 618143</i>
PIGT	100%	100%	<i>?Paroxysmal nocturnal hemoglobinuria 2, 615399</i> <i>Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398</i>
PIGU	100%	100%	<i>Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590</i>
PIGV	100%	100%	<i>Hyperphosphatasia with impaired intellectual development syndrome 1, 239300</i>
PIGW	100%	100%	<i>Glycosylphosphatidylinositol biosynthesis defect 11, 616025</i>
PIGY	100%	100%	<i>Hyperphosphatasia with impaired intellectual development syndrome 6, 616809</i>
PIK3CA	100%	100%	<i>CLOVE syndrome, somatic, 612918</i> <i>Hepatocellular carcinoma, somatic, 114550</i> <i>Breast cancer, somatic, 114480</i> <i>Cerebral cavernous malformations 4, somatic, 619538</i> <i>Ovarian cancer, somatic, 167000</i> <i>Colorectal cancer, somatic, 114500</i> <i>Macrodactyly, somatic, 155500</i> <i>CLAPO syndrome, somatic, 613089</i> <i>Keratosis, seborrhic, somatic, 182000</i> <i>Nevus, epidermal, somatic, 162900</i>

			<i>Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108</i>
<i>PIK3R2</i>	<i>100%</i>	<i>100%</i>	<i>Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387</i>
<i>PISD</i>	<i>100%</i>	<i>100%</i>	<i>Liberfarb syndrome, 618889</i>
<i>PITRM1</i>	<i>100%</i>	<i>100%</i>	<i>Spinocerebellar ataxia, autosomal recessive 30, 619405</i>
<i>PJA1</i>	<i>100%</i>	<i>100%</i>	<i>No OMIM disease ID</i>
<i>PLA2G6</i>	<i>100%</i>	<i>100%</i>	<i>Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600</i>
<i>PLAA</i>	<i>100%</i>	<i>100%</i>	<i>Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527</i>
<i>PLCB1</i>	<i>100%</i>	<i>100%</i>	<i>Developmental and epileptic encephalopathy 12, 613722</i>
<i>PLK1</i>	<i>100%</i>	<i>100%</i>	<i>No OMIM disease ID</i>
<i>PLK4</i>	<i>100%</i>	<i>100%</i>	<i>Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171</i>
<i>PLP1</i>	<i>100%</i>	<i>99%</i>	<i>Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920</i>
<i>PLPBP</i>	<i>100%</i>	<i>100%</i>	<i>Epilepsy, early-onset, vitamin B6-dependent, 617290</i>
<i>PLXNA1</i>	<i>100%</i>	<i>100%</i>	<i>Dworschak-Punetha neurodevelopmental syndrome, 619955</i>
<i>PLXNA2</i>	<i>100%</i>	<i>100%</i>	<i>No OMIM disease ID</i>
<i>PLXND1</i>	<i>100%</i>	<i>100%</i>	<i>No OMIM disease ID</i>
<i>PMM2</i>	<i>100%</i>	<i>100%</i>	<i>Congenital disorder of glycosylation, type Ia, 212065</i>
<i>PMPCA</i>	<i>100%</i>	<i>100%</i>	<i>Spinocerebellar ataxia, autosomal recessive 2, 213200</i>
<i>PMPCB</i>	<i>100%</i>	<i>100%</i>	<i>Multiple mitochondrial dysfunctions syndrome 6, 617954</i>
<i>PNKP</i>	<i>100%</i>	<i>100%</i>	<i>?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402</i>
<i>PNP</i>	<i>100%</i>	<i>100%</i>	<i>Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179</i>
<i>PNPLA6</i>	<i>100%</i>	<i>100%</i>	<i>Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470</i>
<i>POGZ</i>	<i>100%</i>	<i>100%</i>	<i>White-Sutton syndrome, 616364</i>
<i>POLA1</i>	<i>100%</i>	<i>99%</i>	<i>Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030</i>

POLG	100%	100%	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%	83%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR2A	100%	100%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100%	100%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLRMT	100%	100%	Combined oxidative phosphorylation deficiency 55, 619743
POMGNT1	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100%	100%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	100%	100%	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 impaired intellectual development), type B, 1, 613155
POMT2	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156
PORCN	100%	100%	Focal dermal hypoplasia, 305600
POU1F1	100%	100%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU3F3	100%	98%	Snijders Blok-Fisher syndrome, 618604
PPIL1	100%	100%	Pontocerebellar hypoplasia, type 14, 619301
PPM1D	100%	100%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506

PPP1R12A	100%	99%	<i>Genitourinary and/or/brain malformation syndrome, 618820</i>
PPP1R15B	100%	100%	<i>Microcephaly, short stature, and impaired glucose metabolism 2, 616817</i>
PPP1R21	100%	100%	<i>Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383</i>
PPP2CA	100%	100%	<i>Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354</i>
PPP2R1A	94%	94%	<i>Intellectual developmental disorder, autosomal dominant 36, 616362</i>
PPP2R3C	100%	100%	<i>Spermatogenic failure 36, 618420</i> <i>Myoectodermal gonadal dysgenesis syndrome, 618419</i>
PPP2R5B	100%	100%	<i>No OMIM disease ID</i>
PPP2R5C	100%	100%	<i>No OMIM disease ID</i>
PPP2R5D	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 35, 616355</i>
PPP3CA	100%	100%	<i>Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265</i> <i>Developmental and epileptic encephalopathy 91, 617711</i>
PPT1	90%	90%	<i>Ceroid lipofuscinosis, neuronal, 1, 256730</i>
PQBP1	100%	100%	<i>Renpenning syndrome, 309500</i>
PRDM13	100%	100%	<i>Pontocerebellar hypoplasia, type 17, 619909</i> <i>Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761</i>
PRDM15	100%	100%	<i>No OMIM disease ID</i>
PRICKLE2	100%	100%	<i>No OMIM disease ID</i>
PRKACB	100%	99%	<i>Cardioacrofacial dysplasia 2, 619143</i>
PRKAR1A	100%	100%	<i>Pigmented nodular adrenocortical disease, primary, 1, 610489</i> <i>Acrodysostosis 1, with or without hormone resistance, 101800</i> <i>Carney complex, type 1, 160980</i> <i>Myxoma, intracardiac, 255960</i> <i>Adrenocortical tumor, somatic,</i>
PRKAR1B	100%	100%	<i>Marbach-Schaaf neurodevelopmental syndrome, 619680</i>
PRMT7	100%	100%	<i>Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157</i>
PRODH	100%	100%	<i>Hyperprolinemia, type I, 239500</i>
PRPF8	100%	100%	<i>Retinitis pigmentosa 13, 600059</i>
PRPS1	100%	100%	<i>Arts syndrome, 301835</i> <i>Phosphoribosylpyrophosphate synthetase superactivity, 300661</i> <i>Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070</i> <i>Deafness, X-linked 1, 304500</i> <i>Gout, PRPS-related, 300661</i>
PRR12	100%	100%	<i>Neuroocular syndrome, 619539</i>
PRSS12	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 1, 249500</i>

<i>PRUNE1</i>	93%	93%	<i>Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481</i>
<i>PSAP</i>	100%	100%	<i>Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539</i>
<i>PSAT1</i>	100%	100%	<i>Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992</i>
<i>PSMC5</i>	100%	100%	<i>No OMIM disease ID</i>
<i>PSMD12</i>	100%	100%	<i>Stankiewicz-Isidor syndrome, 617516</i>
<i>PSPH</i>	100%	100%	<i>Phosphoserine phosphatase deficiency, 614023</i>
<i>PTCH1</i>	100%	100%	<i>Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400</i>
<i>PTCHD1</i>	100%	100%	<i>No OMIM disease ID</i>
<i>PTDSS1</i>	100%	100%	<i>Lenz-Majewski hyperostotic dwarfism, 151050</i>
<i>PTEN</i>	100%	100%	<i>Cowden syndrome 1, 158350 Lhermitte-Duclos disease, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309</i>
<i>PTF1A</i>	100%	100%	<i>Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935</i>
<i>PTPN11</i>	100%	100%	<i>Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785</i>
<i>PTPN23</i>	100%	100%	<i>Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890</i>
<i>PTRH2</i>	100%	100%	<i>Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263</i>
<i>PTRHD1</i>	100%	100%	<i>No OMIM disease ID</i>
<i>PTS</i>	100%	100%	<i>Hyperphenylalaninemia, BH4-deficient, A, 261640</i>
<i>PUF60</i>	100%	100%	<i>Verheij syndrome, 615583</i>
<i>PUM1</i>	100%	100%	<i>Spinocerebellar ataxia 47, 617931</i>
<i>PURA</i>	100%	100%	<i>Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158</i>
<i>PUS1</i>	100%	100%	<i>Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462</i>
<i>PUS3</i>	100%	100%	<i>Neurodevelopmental disorder with microcephaly and gray sclerae, 617051</i>
<i>PUS7</i>	100%	100%	<i>Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342</i>

PYCR1	100%	100%	<i>Cutis laxa, autosomal recessive, type IIB, 614438</i> <i>Cutis laxa, autosomal recessive, type IIB, 612940</i>
PYCR2	100%	100%	<i>Leukodystrophy, hypomyelinating, 10, 616420</i>
QARS1	100%	100%	<i>Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760</i>
QDPR	100%	100%	<i>Hyperphenylalaninemia, BH4-deficient, C, 261630</i>
QRICH1	100%	100%	<i>Ververi-Brady syndrome, 617982</i>
RAB11B	100%	100%	<i>Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807</i>
RAB14	100%	100%	<i>No OMIM disease ID</i>
RAB18	100%	100%	<i>Warburg micro syndrome 3, 614222</i>
RAB23	100%	100%	<i>Carpenter syndrome, 201000</i>
RAB27A	100%	100%	<i>Griscelli syndrome, type 2, 607624</i>
RAB39B	100%	100%	<i>Intellectual developmental disorder, X-linked 72, 300271</i> <i>Waisman syndrome, 311510</i>
RAB3GAP1	99%	99%	<i>Martsolf syndrome 2, 619420</i> <i>Warburg micro syndrome 1, 600118</i>
RAB3GAP2	100%	100%	<i>Martsolf syndrome 1, 212720</i> <i>Warburg micro syndrome 2, 614225</i>
RAC1	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 48, 617751</i>
RAC3	100%	100%	<i>Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577</i>
RAD21	100%	100%	<i>Cornelia de Lange syndrome 4, 614701</i> <i>?Mungan syndrome, 611376</i>
RAF1	100%	100%	<i>Cardiomyopathy, dilated, 1NN, 615916</i> <i>Noonan syndrome 5, 611553</i> <i>LEOPARD syndrome 2, 611554</i>
RAI1	100%	100%	<i>Smith-Magenis syndrome, 182290</i>
RALA	100%	100%	<i>Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311</i>
RALGAPA1	100%	100%	<i>Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797</i>
RARB	100%	100%	<i>Microphthalmia, syndromic 12, 615524</i>
RARS1	94%	94%	<i>Leukodystrophy, hypomyelinating, 9, 616140</i>
RARS2	100%	100%	<i>Pontocerebellar hypoplasia, type 6, 611523</i>
RBBP8	100%	100%	<i>Seckel syndrome 2, 606744</i> <i>Jawad syndrome, 251255</i> <i>Pancreatic carcinoma, somatic,</i>
RBFOX1	100%	100%	<i>No OMIM disease ID</i>
RBM10	100%	100%	<i>TARP syndrome, 311900</i>
RBM28	100%	100%	<i>?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079</i>

RBPJ	100%	100%	Adams-Oliver syndrome 3, 614814
RCBTB1	100%	100%	Retinal dystrophy with or without extraocular anomalies, 617175
RECQL4	100%	100%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
RELN	100%	100%	Lissencephaly 2 (Norman-Roberts type), 257320
RERE	100%	100%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	98%	98%	No OMIM disease ID
RFT1	100%	100%	Congenital disorder of glycosylation, type In, 612015
RFX3	100%	99%	No OMIM disease ID
RFX4	100%	100%	No OMIM disease ID
RFX7	100%	100%	No OMIM disease ID
RHEB	100%	100%	No OMIM disease ID
RHOBTB2	100%	100%	Developmental and epileptic encephalopathy 64, 618004
RIC1	100%	100%	CATIFA syndrome, 618761
RIMS2	100%	100%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIT1	100%	100%	Noonan syndrome 8, 615355
RLIM	100%	100%	Tonne-Kalscheuer syndrome, 300978
RMND1	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH2A	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91%	91%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	100%	100%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100%	100%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	100%	100%	Tenorio syndrome, 616260
RNF13	100%	100%	Developmental and epileptic encephalopathy 73, 618379
RNF2	100%	100%	Luo-Schoch-Yamamoto syndrome, 619460
RNF220	100%	100%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNPC3	100%	100%	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%	100%	<i>Kohlschutter-Tonz syndrome, 226750</i>
ROR2	100%	100%	<i>Brachydactyly, type B1, 113000</i> <i>Robinow syndrome, autosomal recessive, 268310</i>
RORA	100%	100%	<i>Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060</i>
RORB	100%	100%	<i>No OMIM disease ID</i>
RPGRIP1L	100%	100%	<i>Joubert syndrome 7, 611560</i> <i>Meckel syndrome 5, 611561</i> <i>?COACH syndrome 3, 619113</i>
RPIA	100%	100%	<i>Ribose 5-phosphate isomerase deficiency, 608611</i>
RPL10	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic 35, 300998</i>
RPS19	100%	100%	<i>Diamond-Blackfan anemia 1, 105650</i>
RPS6KA3	100%	100%	<i>Intellectual developmental disorder, X-linked 19, 300844</i> <i>Coffin-Lowry syndrome, 303600</i>
RRM2B	100%	100%	<i>Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075</i> <i>Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075</i> <i>Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315</i> <i>Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077</i>
RRP7A	100%	100%	<i>?Microcephaly 28, primary, autosomal recessive, 619453</i>
RSPRY1	100%	100%	<i>Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723</i>
RSRC1	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 70, 618402</i>
RTEL1	100%	100%	<i>Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373</i> <i>Dyskeratosis congenita, autosomal dominant 4, 615190</i> <i>Dyskeratosis congenita, autosomal recessive 5, 615190</i>
RTN4IP1	100%	100%	<i>Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732</i>
RTTN	100%	100%	<i>Microcephaly, short stature, and polymicrogyria with seizures, 614833</i>
RUBCN	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 15, 615705</i>
RUSC2	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 61, 617773</i>
RXYLT1	100%	100%	<i>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041</i>
SALL1	100%	100%	<i>Townes-Brocks syndrome 1, 107480</i> <i>Townes-Brocks branchiootorenal-like syndrome, 107480</i>
SAMD9	100%	100%	<i>Tumoral calcinosis, familial, normophosphatemic, 610455</i> <i>Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041</i> <i>MIRAGE syndrome, 617053</i>
SAMHD1	100%	100%	<i>?Chilblain lupus 2, 614415</i> <i>Aicardi-Goutieres syndrome 5, 612952</i>
SARS1	100%	100%	<i>Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709</i>

SATB1	100%	100%	<i>Kohlschutter-Tonz syndrome-like, 619229</i> <i>Developmental delay with dysmorphic facies and dental anomalies, 619228</i>
SATB2	100%	100%	<i>Glass syndrome, 612313</i>
SBDS	100%	100%	<i>Shwachman-Diamond syndrome 1, 260400</i>
SC5D	100%	100%	<i>Lathosterolosis, 607330</i>
SCAF4	100%	100%	<i>No OMIM disease ID</i>
SCAMP5	100%	100%	<i>No OMIM disease ID</i>
SCAPER	100%	100%	<i>Intellectual developmental disorder and retinitis pigmentosa, 618195</i>
SCN1A	100%	100%	<i>Developmental and epileptic encephalopathy 6B, non-Dravet, 619317</i> <i>Migraine, familial hemiplegic, 3, 609634</i> <i>Dravet syndrome, 607208</i> <i>Febrile seizures, familial, 3A, 604403</i> <i>Generalized epilepsy with febrile seizures plus, type 2, 604403</i>
SCN1B	100%	100%	<i>Generalized epilepsy with febrile seizures plus, type 1, 604233</i> <i>Developmental and epileptic encephalopathy 52, 617350</i> <i>Cardiac conduction defect, nonspecific, 612838</i> <i>Atrial fibrillation, familial, 13, 615377</i> <i>Brugada syndrome 5, 612838</i>
SCN2A	100%	100%	<i>Seizures, benign familial infantile, 3, 607745</i> <i>Developmental and epileptic encephalopathy 11, 613721</i> <i>Episodic ataxia, type 9, 618924</i>
SCN3A	100%	100%	<i>Epilepsy, familial focal, with variable foci 4, 617935</i> <i>Developmental and epileptic encephalopathy 62, 617938</i>
SCN8A	100%	100%	<i>?Myoclonus, familial, 2, 618364</i> <i>Seizures, benign familial infantile, 5, 617080</i> <i>Cognitive impairment with or without cerebellar ataxia, 614306</i> <i>Developmental and epileptic encephalopathy 13, 614558</i>
SCO1	100%	100%	<i>Mitochondrial complex IV deficiency, nuclear type 4, 619048</i>
SCO2	100%	100%	<i>Myopia 6, 608908</i> <i>Mitochondrial complex IV deficiency, nuclear type 2, 604377</i>
SCUBE3	100%	100%	<i>Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184</i>
SCYL1	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 21, 616719</i>
SDCCAG8	100%	100%	<i>Senior-Loken syndrome 7, 613615</i> <i>Bardet-Biedl syndrome 16, 615993</i>
SDHA	100%	100%	<i>Cardiomyopathy, dilated, 1GG, 613642</i> <i>Mitochondrial complex II deficiency, nuclear type 1, 252011</i>

			<i>Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165</i>
<i>SEC31A</i>	100%	100%	<i>?Halperin-Birk syndrome, 618651</i>
<i>SEMA3E</i>	100%	100%	<i>No OMIM disease ID</i>
<i>SEPSECS</i>	100%	100%	<i>Pontocerebellar hypoplasia type 2D, 613811</i>
<i>SERAC1</i>	100%	100%	<i>3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739</i>
<i>SET</i>	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 58, 618106</i>
<i>SETBP1</i>	100%	100%	<i>Schinzel-Giedion midface retraction syndrome, 269150 Intellectual developmental disorder, autosomal dominant 29, 616078</i>
<i>SETD1A</i>	100%	100%	<i>Epilepsy, early-onset, with or without developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056</i>
<i>SETD1B</i>	100%	100%	<i>Intellectual developmental disorder with seizures and language delay, 619000</i>
<i>SETD2</i>	100%	100%	<i>Luscan-Lumish syndrome, 616831</i>
<i>SETD5</i>	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 23, 615761</i>
<i>SFXN4</i>	100%	100%	<i>Combined oxidative phosphorylation deficiency 18, 615578</i>
<i>SGPL1</i>	100%	100%	<i>Nephrotic syndrome, type 14, 617575</i>
<i>SGSH</i>	100%	100%	<i>Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900</i>
<i>SHANK1</i>	100%	100%	<i>No OMIM disease ID</i>
<i>SHANK2</i>	100%	100%	<i>No OMIM disease ID</i>
<i>SHANK3</i>	100%	99%	<i>Phelan-McDermid syndrome, 606232</i>
<i>SHH</i>	100%	100%	<i>Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945</i>
<i>SHMT2</i>	100%	100%	<i>Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121</i>
<i>SHOC2</i>	100%	100%	<i>Noonan syndrome-like with loose anagen hair 1, 607721</i>
<i>SHROOM4</i>	100%	100%	<i>No OMIM disease ID</i>
<i>SIAH1</i>	100%	100%	<i>Buratti-Harel syndrome, 619314</i>
<i>SIK1</i>	100%	100%	<i>Developmental and epileptic encephalopathy 30, 616341</i>
<i>SIL1</i>	100%	100%	<i>Marinesco-Sjogren syndrome, 248800</i>
<i>SIN3A</i>	100%	100%	<i>Witteveen-Kolk syndrome, 613406</i>
<i>SIN3B</i>	100%	100%	<i>No OMIM disease ID</i>
<i>SIX3</i>	100%	100%	<i>Schizencephaly, 269160 Holoprosencephaly 2, 157170</i>
<i>SKI</i>	100%	100%	<i>Shprintzen-Goldberg syndrome, 182212</i>

TTC37	100%	100%	Trichohepatoenteric syndrome 1, 222470
SLC12A2	100%	100%	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081
SLC12A5	100%	100%	Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000 Charcot-Marie-Tooth disease, axonal, type 2II, 620068
SLC13A5	100%	100%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A2	100%	100%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	100%	100%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	100%	98%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	100%	100%	Dicarboxylic aminoaciduria, 222730
SLC1A2	100%	100%	Developmental and epileptic encephalopathy 41, 617105
SLC1A4	100%	100%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	100%	100%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A12	100%	100%	Developmental and epileptic encephalopathy 39, 612949
SLC25A15	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100%	100%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	100%	100%	Fontaine progeroid syndrome, 612289
SLC25A42	100%	100%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC2A1	100%	100%	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%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	100%	100%	Congenital disorder of glycosylation, type IIj, 603585
SLC35A2	100%	100%	Congenital disorder of glycosylation, type IIk, 300896
SLC35A3	98%	93%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100%	100%	No OMIM disease ID
SLC35C1	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC38A3	100%	100%	Developmental and epileptic encephalopathy 102, 619881
SLC39A14	94%	94%	?Hyperostosis cranialis interna, 144755 Hypermanganesemia with dystonia 2, 617013

SLC39A8	100%	100%	<i>Congenital disorder of glycosylation, type II n, 616721</i>
SLC45A1	100%	100%	<i>Intellectual developmental disorder with neuropsychiatric features, 617532</i>
SLC46A1	100%	100%	<i>Folate malabsorption, hereditary, 229050</i>
SLC4A4	100%	100%	<i>Renal tubular acidosis, proximal, with ocular abnormalities, 604278</i>
SLC5A6	100%	100%	<i>Sodium-dependent multivitamin transporter deficiency, 618973</i> <i>Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903</i>
SLC6A1	100%	100%	<i>Myoclonic-atonic epilepsy, 616421</i>
SLC6A17	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 48, 616269</i>
SLC6A19	100%	100%	<i>Iminoglycinuria, digenic, 242600</i> <i>Hartnup disorder, 234500</i> <i>Hyperglycinuria, 138500</i>
SLC6A3	100%	100%	<i>Parkinsonism-dystonia, infantile, 1, 613135</i>
SLC6A8	100%	100%	<i>Cerebral creatine deficiency syndrome 1, 300352</i>
SLC6A9	100%	100%	<i>Glycine encephalopathy with normal serum glycine, 617301</i>
SLC7A7	100%	100%	<i>Lysinuric protein intolerance, 222700</i>
SLC9A6	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243</i>
SLC9A7	100%	100%	<i>Intellectual developmental disorder, X-linked 108, 301024</i>
SMAD4	100%	100%	<i>Pancreatic cancer, somatic, 260350</i> <i>Myhre syndrome, 139210</i> <i>Polyposis, juvenile intestinal, 174900</i> <i>Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050</i>
SMARCA1	100%	100%	<i>No OMIM disease ID</i>
SMARCA2	100%	100%	<i>Nicolaidis-Baraitser syndrome, 601358</i> <i>Blepharophimosis-impaired intellectual development syndrome, 619293</i>
SMARCA4	100%	100%	<i>Coffin-Siris syndrome 4, 614609</i>
SMARCA5	100%	100%	<i>No OMIM disease ID</i>
SMARCB1	100%	100%	<i>Rhabdoid tumors, somatic, 609322</i> <i>Coffin-Siris syndrome 3, 614608</i>
SMARCC2	100%	100%	<i>Coffin-Siris syndrome 8, 618362</i>
SMARCD1	100%	100%	<i>Coffin-Siris syndrome 11, 618779</i>
SMARCE1	100%	100%	<i>Coffin-Siris syndrome 5, 616938</i>
SMC1A	100%	100%	<i>Cornelia de Lange syndrome 2, 300590</i> <i>Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044</i>
SMC3	100%	100%	<i>Cornelia de Lange syndrome 3, 610759</i>
SMG8	100%	100%	<i>Alzahrani-Kuwahara syndrome, 619268</i>

SMG9	100%	100%	<i>Heart and brain malformation syndrome, 616920</i> <i>Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995</i>
SMOC1	100%	100%	<i>Microphthalmia with limb anomalies, 206920</i>
SMPD1	100%	100%	<i>Niemann-Pick disease, type B, 607616</i> <i>Niemann-Pick disease, type A, 257200</i>
SMPD4	100%	100%	<i>Neurodevelopmental disorder with microcephaly, arthrogyriposis, and structural brain anomalies, 618622</i>
SMS	100%	99%	<i>Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583</i>
SNAP25	100%	100%	<i>?Myasthenic syndrome, congenital, 18, 616330</i>
SNAP29	100%	100%	<i>Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528</i>
SNIP1	100%	100%	<i>Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501</i>
SNORD118	NC	NC	<i>Leukoencephalopathy, brain calcifications, and cysts, 614561</i>
SNRPB	100%	100%	<i>Cerebrocostomandibular syndrome, 117650</i>
SNRPN	100%	100%	<i>No OMIM disease ID</i>
SNX14	100%	100%	<i>Spinocerebellar ataxia, autosomal recessive 20, 616354</i>
SNX27	100%	100%	<i>No OMIM disease ID</i>
SOBP	100%	100%	<i>Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671</i>
SON	100%	100%	<i>ZTTK syndrome, 617140</i>
SOS1	100%	100%	<i>Noonan syndrome 4, 610733</i> <i>?Fibromatosis, gingival, 1, 135300</i>
SOS2	100%	100%	<i>Noonan syndrome 9, 616559</i>
SOX10	100%	100%	<i>Waardenburg syndrome, type 4C, 613266</i> <i>PCWH syndrome, 609136</i> <i>Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584</i>
SOX11	100%	100%	<i>Coffin-Siris syndrome 9, 615866</i>
SOX2	100%	100%	<i>Optic nerve hypoplasia and abnormalities of the central nervous system, 206900</i> <i>Microphthalmia, syndromic 3, 206900</i>
SOX3	100%	100%	<i>Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123</i> <i>Panhypopituitarism, X-linked, 312000</i>
SOX4	100%	100%	<i>Coffin-Siris syndrome 10, 618506</i>
SOX5	100%	100%	<i>Lamb-Shaffer syndrome, 616803</i>
SOX6	100%	99%	<i>Tolchin-Le Caignec syndrome, 618971</i>
SPART	100%	100%	<i>Troyer syndrome, 275900</i>
SPAST	100%	100%	<i>Spastic paraplegia 4, autosomal dominant, 182601</i>
SPATA5	100%	100%	<i>Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577</i>

SPATA5L1	100%	100%	Deafness, autosomal recessive 119, 619615 Neurodevelopmental disorder with hearing loss and spasticity, 619616
SPECC1L	100%	100%	Teebi hypertelorism syndrome 1, 145420 ?Facial clefting, oblique, 1, 600251
SPEN	100%	100%	Radio-Tartaglia syndrome, 619312
SPG11	100%	100%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPOCK1	100%	100%	No OMIM disease ID
SPOP	100%	100%	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829
SPR	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100%	100%	Legius syndrome, 611431
SPRED2	100%	100%	Noonan syndrome 14, 619745
SPTAN1	100%	100%	Developmental and epileptic encephalopathy 5, 613477
SPTBN1	100%	100%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100%	100%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100%	100%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SRCAP	100%	100%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140
SRD5A3	100%	100%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SRP54	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRPX2	100%	100%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM2	100%	100%	No OMIM disease ID
SSR4	100%	100%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	97%	95%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98%	98%	Salt and pepper developmental regression syndrome, 609056
STAG1	100%	100%	Intellectual developmental disorder, autosomal dominant 47, 617635
STAG2	100%	100%	Holoprosencephaly 13, X-linked, 301043 Mullegama-Klein-Martinez syndrome, 301022
STAMBP	100%	100%	Microcephaly-capillary malformation syndrome, 614261
CXorf56	100%	100%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	100%	100%	Microcephaly 7, primary, autosomal recessive, 612703

STRA6	100%	100%	<i>Microphthalmia, syndromic 9, 601186</i> <i>Microphthalmia, isolated, with coloboma 8, 601186</i>
STRADA	100%	100%	<i>Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087</i>
STT3A	100%	100%	<i>Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714</i> <i>Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596</i>
STT3B	100%	100%	<i>Congenital disorder of glycosylation, type Ix, 615597</i>
STX1B	100%	100%	<i>Generalized epilepsy with febrile seizures plus, type 9, 616172</i>
STXBP1	100%	100%	<i>Developmental and epileptic encephalopathy 4, 612164</i>
SUCLA2	100%	100%	<i>Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073</i>
SUCLG1	100%	100%	<i>Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400</i>
SUFU	100%	100%	<i>Joubert syndrome 32, 617757</i> <i>Basal cell nevus syndrome, 109400</i>
SUMF1	100%	100%	<i>Multiple sulfatase deficiency, 272200</i>
SUOX	100%	100%	<i>Sulfite oxidase deficiency, 272300</i>
SUPT16H	100%	100%	<i>Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480</i>
SURF1	100%	100%	<i>Charcot-Marie-Tooth disease, type 4K, 616684</i> <i>Mitochondrial complex IV deficiency, nuclear type 1, 220110</i>
SUZ12	100%	100%	<i>Imagawa-Matsumoto syndrome, 618786</i>
SVBP	100%	100%	<i>Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569</i>
SYN1	100%	100%	<i>Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491</i> <i>Intellectual developmental disorder, X-linked 50, 300115</i>
SYNCRIP	100%	100%	<i>No OMIM disease ID</i>
SYNGAP1	100%	100%	<i>Intellectual developmental disorder, autosomal dominant 5, 612621</i>
SYNJ1	100%	100%	<i>Parkinson disease 20, early-onset, 615530</i> <i>Developmental and epileptic encephalopathy 53, 617389</i>
SYP	100%	100%	<i>Intellectual developmental disorder, X-linked 96, 300802</i>
SYT1	100%	100%	<i>Baker-Gordon syndrome, 618218</i>
SZT2	100%	100%	<i>Developmental and epileptic encephalopathy 18, 615476</i>
TACO1	100%	100%	<i>Mitochondrial complex IV deficiency, nuclear type 8, 619052</i>
TAF1	100%	100%	<i>Intellectual developmental disorder, X-linked syndromic 33, 300966</i> <i>Dystonia-Parkinsonism, X-linked, 314250</i>
TAF13	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 60, 617432</i>
TAF1C	100%	100%	<i>No OMIM disease ID</i>
TAF2	100%	100%	<i>Intellectual developmental disorder, autosomal recessive 40, 615599</i>
TAF4	90%	85%	<i>No OMIM disease ID</i>

TAF6	100%	100%	Alazami-Yuan syndrome, 617126
TAF8	89%	89%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972
TANC2	100%	100%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100%	100%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100%	100%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TASP1	100%	100%	Suleiman-El-Hattab syndrome, 618950
TAT	100%	100%	Tyrosinemia, type II, 276600
TBC1D20	100%	100%	Warburg micro syndrome 4, 615663
TBC1D23	100%	100%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100%	100%	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with paroxysmal exercise-induced 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	100%	100%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D7	100%	100%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	100%	100%	Intellectual developmental disorder, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBP	100%	100%	Spinocerebellar ataxia 17, 607136
TBR1	100%	100%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	98%	96%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TCF20	100%	100%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF4	100%	100%	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	100%	100%	No OMIM disease ID
TCN2	100%	100%	Transcobalamin II deficiency, 275350

TCTN2	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDP2	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100%	100%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	100%	100%	Intellectual developmental disorder, autosomal recessive 14, 614020
TELO2	100%	100%	You-Hoover-Fong syndrome, 616954
TENM3	100%	100%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TET3	100%	100%	Beck-Fahrner syndrome, 618798
TFAP2A	100%	100%	Branchiooculofacial syndrome, 113620
TFE3	100%	100%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 Renal cell carcinoma, papillary, 1, 300854
TGDS	100%	100%	Catel-Manzke syndrome, 616145
TGFBR1	100%	100%	Loeys-Dietz syndrome 1, 609192
TGIF1	100%	100%	Holoprosencephaly 4, 142946
TH	100%	100%	Segawa syndrome, recessive, 605407
THOC2	100%	100%	Intellectual developmental disorder, X-linked 12, 300957
THOC6	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100%	100%	Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650
THUMPD1	100%	100%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989
TIAM1	100%	100%	Neurodevelopmental disorder with language delay and seizures, 619908
TIMM50	100%	100%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TINF2	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TKFC	100%	100%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98%	98%	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	100%	100%	Intellectual developmental disorder, autosomal dominant 57, 618050
TMCO1	88%	88%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMEM106B	100%	100%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	100%	100%	Congenital disorder of glycosylation, type IIk, 614727

TMEM216	100%	100%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM222	100%	100%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	100%	100%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100%	100%	Joubert syndrome 14, 614424
TMEM240	100%	100%	Spinocerebellar ataxia 21, 607454
TMEM63A	100%	100%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM63C	100%	100%	Spastic paraplegia 87, autosomal recessive, 619966
TMEM67	100%	98%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TMEM70	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100%	100%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMLHE	100%	99%	No OMIM disease ID
TMTC3	100%	100%	Lissencephaly 8, 617255
TMX2	100%	100%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNIK	100%	100%	Intellectual developmental disorder, autosomal recessive 54, 617028
TNPO2	100%	100%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNR	100%	100%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNRC6B	100%	100%	Global developmental delay with speech and behavioral abnormalities, 619243
TOE1	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100%	100%	Joubert syndrome 37, 619185
TOMM70	100%	100%	No OMIM disease ID
TOR1A	91%	91%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TP53RK	100%	100%	Galloway-Mowat syndrome 4, 617730
TP73	100%	100%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPI1	100%	100%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	100%	100%	Thyroid dysmorphogenesis 2A, 274500
TPP1	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100%	100%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220

TPRKB	82%	81%	Galloway-Mowat syndrome 5, 617731
TRAF7	100%	100%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100%	100%	Seckel syndrome 9, 616777
TRAK1	100%	100%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2L	100%	100%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	100%	100%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100%	100%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	100%	Intellectual developmental disorder, autosomal recessive 13, 613192
TREX1	100%	100%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM8	100%	100%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	100%	100%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIP12	100%	100%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIT1	100%	100%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100%	100%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM3	100%	100%	No OMIM disease ID
TRRAP	100%	100%	?Deafness, autosomal dominant 75, 618778 Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100%	100%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioliomyomatosis, 606690
TSC2	100%	100%	Lymphangioliomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TSEN15	100%	100%	Pontocerebellar hypoplasia, type 2F, 617026

TSEN2	100%	100%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100%	100%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSMF	94%	94%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100%	100%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	99%	99%	Intellectual developmental disorder, X-linked 58, 300210
TTC19	100%	100%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC5	100%	100%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC8	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100%	100%	Intellectual developmental disorder, autosomal recessive 39, 615541
TUBA1A	100%	100%	Lissencephaly 3, 611603
TUBA8	100%	100%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840
TUBB	100%	99%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB2A	100%	100%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	100%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	99%	96%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	97%	97%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	100%	100%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST1	100%	100%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWINK	100%	100%	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%	100%	No OMIM disease ID
UBA5	100%	100%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE2A	100%	100%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860
UBE3A	100%	100%	Angelman syndrome, 105830
UBE3B	100%	100%	Kaufman oculocerebrofacial syndrome, 244450
UBE4A	100%	100%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBR1	98%	98%	Johanson-Blizzard syndrome, 243800
UBR7	100%	100%	Li-Campeau syndrome, 619189
UBTF	100%	100%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	100%	100%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100%	100%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100%	100%	?Hip dysplasia, Beukes type, 142669 Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 Developmental and epileptic encephalopathy 106, 620028
UGDH	100%	100%	Developmental and epileptic encephalopathy 84, 618792
UGP2	96%	94%	Developmental and epileptic encephalopathy 83, 618744
UNC13A	100%	100%	No OMIM disease ID
UNC45A	100%	100%	Osteotohepatoenteric syndrome, 619377
UNC80	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF1	100%	99%	No OMIM disease ID
UPF3B	100%	100%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UROC1	100%	100%	?Urocanase deficiency, 276880
USP27X	100%	100%	Intellectual developmental disorder, X-linked 105, 300984
USP7	100%	100%	Hao-Fountain syndrome, 616863
USP9X	100%	100%	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
VAMP1	100%	100%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100%	100%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VARS1	100%	100%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	100%	Combined oxidative phosphorylation deficiency 20, 615917
VLDLR	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050

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

WFS1	100%	100%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WNK3	100%	100%	No OMIM disease ID
WWOX	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XPA	100%	100%	Xeroderma pigmentosum, group A, 278700
XRCC4	100%	100%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100%	100%	Desbuquois dysplasia 2, 615777
YIF1B	90%	90%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100%	100%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	100%	100%	?Optic atrophy 11, 617302
YWHAE	100%	100%	No OMIM disease ID
YWHAG	100%	100%	Developmental and epileptic encephalopathy 56, 617665
YY1	100%	100%	Gabriele-de Vries syndrome, 617557
ZBTB11	100%	100%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type,
ZBTB18	100%	100%	Intellectual developmental disorder, autosomal dominant 22, 612337
ZBTB20	100%	100%	Primrose syndrome, 259050
ZBTB24	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB7A	100%	100%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769
ZC3H14	100%	100%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZDHHHC9	100%	100%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799
ZEB2	97%	97%	Mowat-Wilson syndrome, 235730
ZFHX4	100%	99%	No OMIM disease ID
ZFYVE26	100%	100%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	100%	100%	?Craniosynostosis 6, 616602 Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	100%	Holoprosencephaly 5, 609637
ZMIZ1	100%	100%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYM2	100%	100%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522

ZMYND11	100%	100%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZNF142	100%	100%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	100%	100%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99%	99%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100%	100%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	100%	100%	SIMHA syndrome, 619557
ZNF41	100%	100%	No OMIM disease ID
ZNF462	100%	100%	Weiss-Kruszka syndrome, 618619
ZNF526	100%	100%	Dentici-Novelli neurodevelopmental syndrome, 619877
ZNF699	100%	100%	DEGCAGS syndrome, 619488
ZNF711	100%	100%	Intellectual developmental disorder, X-linked 97, 300803
ZSWIM6	98%	96%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 Acromelic frontonasal dysostosis, 603671

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

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

TWIST X2 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 : November 28th , 2022.

This list is accurate for panel version DG 3.5.0

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