

INTELLECTUAL DISABILITY GENE PANEL DG 3.4.0 (1612 genes)

Releasedate: 19-04-2022

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

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

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

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

| | | | |
|----------|--------|--------|---|
| ARF3 | 100,0% | 100,0% | No OMIM Disease ID |
| ARFGF1 | 100,0% | 100,0% | No OMIM Disease ID |
| ARFGF2 | 100,0% | 100,0% | Periventricular heterotopia with microcephaly, 608097 |
| ARG1 | 92,9% | 92,9% | Argininemia, 207800 |
| ARHGAP31 | 100,0% | 100,0% | Adams-Oliver syndrome 1, 100300 |
| ARHGAP35 | 100,0% | 100,0% | No OMIM Disease ID |
| ARHGEF6 | 100,0% | 100,0% | No OMIM Disease ID |
| ARHGEF9 | 97,2% | 97,2% | Developmental and epileptic encephalopathy 8, 300607 |
| ARID1A | 100,0% | 100,0% | Coffin-Siris syndrome 2, 614607 |
| ARID1B | 98,6% | 98,3% | Coffin-Siris syndrome 1, 135900 |
| ARID2 | 100,0% | 100,0% | Coffin-Siris syndrome 6, 617808 |
| ARL13B | 100,0% | 100,0% | Joubert syndrome 8, 612291 |
| ARL6 | 100,0% | 100,0% | Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 |
| ARMC9 | 100,0% | 100,0% | Joubert syndrome 30, 617622 |
| ARPC4 | 100,0% | 100,0% | No OMIM Disease ID |
| ARSA | 100,0% | 100,0% | Metachromatic leukodystrophy, 250100 |
| ARSL | 100,0% | 100,0% | Chondrodysplasia punctata, X-linked recessive, 302950 |
| ARV1 | 100,0% | 100,0% | Developmental and epileptic encephalopathy 38, 617020 |
| ARX | 99,0% | 96,8% | Proud syndrome, 300004 Hydranencephaly with abnormal genitalia, 300215 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Lissencephaly, X-linked 2, 300215 Intellectual developmental disorder, X-linked 29, 300419 |
| ASAH1 | 100,0% | 100,0% | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000 |
| ASH1L | 98,7% | 98,7% | Intellectual developmental disorder, autosomal dominant 52, 617796 |
| ASL | 100,0% | 100,0% | Argininosuccinic aciduria, 207900 |
| ASNS | 100,0% | 100,0% | Asparagine synthetase deficiency, 615574 |
| ASPA | 100,0% | 100,0% | Canavan disease, 271900 |
| ASPM | 100,0% | 100,0% | Microcephaly 5, primary, autosomal recessive, 608716 |
| ASS1 | 100,0% | 100,0% | Citrullinemia, 215700 |
| ASXL1 | 99,9% | 99,9% | Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039 |
| ASXL2 | 100,0% | 100,0% | Shashi-Pena syndrome, 617190 |
| ASXL3 | 100,0% | 100,0% | Bainbridge-Ropers syndrome, 615485 |

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

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

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

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

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

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

| | | | |
|---------|--------|--------|--|
| CHRM1 | 100,0% | 100,0% | No OMIM Disease ID |
| CHRNA4 | 100,0% | 100,0% | Epilepsy, nocturnal frontal lobe, 1, 600513 |
| CIC | 100,0% | 100,0% | Intellectual developmental disorder, autosomal dominant 45, 617600 |
| CIT | 100,0% | 100,0% | Microcephaly 17, primary, autosomal recessive, 617090 |
| CKAP2L | 100,0% | 100,0% | Filippi syndrome, 272440 |
| CLCN3 | 96,8% | 96,7% | Neurodevelopmental disorder with seizures and brain abnormalities, 619517 Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 |
| CLCN4 | 100,0% | 100,0% | Raynaud-Claes syndrome, 300114 |
| CLDN11 | 100,0% | 100,0% | Leukodystrophy, hypomyelinating, 22, 619328 |
| CLIC2 | 100,0% | 100,0% | ?Intellectual developmental disorder, X-linked, syndromic 32, 300886 |
| CLIP1 | 100,0% | 100,0% | No OMIM Disease ID |
| CLN3 | 92,7% | 92,5% | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| CLN5 | 71,7% | 71,6% | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 100,0% | 100,0% | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780 |
| CLN8 | 100,0% | 100,0% | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143 |
| CLP1 | 100,0% | 100,0% | Pontocerebellar hypoplasia, type 10, 615803 |
| CLPB | 100,0% | 100,0% | Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 |
| CLTC | 100,0% | 100,0% | Intellectual developmental disorder, autosomal dominant 56, 617854 |
| CNKS2 | 100,0% | 100,0% | Intellectual developmental disorder, X-linked, syndromic, Houge type, 301008 |
| CNNM2 | 100,0% | 100,0% | Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418 |
| CNOT1 | 100,0% | 100,0% | Vissers-Bodmer syndrome, 619033 Holoprosencephaly 12, with or without pancreatic agenesis, 618500 |
| CNOT2 | 100,0% | 100,0% | Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 |
| CNOT3 | 100,0% | 100,0% | Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 |
| CNPY3 | 100,0% | 100,0% | Developmental and epileptic encephalopathy 60, 617929 |
| CNTNAP1 | 100,0% | 100,0% | Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186 |
| CNTNAP2 | 100,0% | 100,0% | Pitt-Hopkins like syndrome 1, 610042 |
| COA8 | 93,5% | 93,5% | Mitochondrial complex IV deficiency, nuclear type 17, 619061 |
| COASY | 100,0% | 100,0% | Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643 |
| COG1 | 100,0% | 100,0% | Congenital disorder of glycosylation, type IIg, 611209 |

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

| | | | |
|----------|--------|--------|---|
| RARS1 | 94,4% | 94,4% | Leukodystrophy, hypomyelinating, 9, 616140 |
| RARS2 | 100,0% | 100,0% | Pontocerebellar hypoplasia, type 6, 611523 |
| RBBP8 | 100,0% | 100,0% | Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic, |
| RBFox1 | 100,0% | 99,7% | No OMIM Disease ID |
| RBM10 | 100,0% | 100,0% | TARP syndrome, 311900 |
| RBM28 | 100,0% | 100,0% | ?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 |
| RBPJ | 100,0% | 100,0% | Adams-Oliver syndrome 3, 614814 |
| RCBTB1 | 100,0% | 100,0% | Retinal dystrophy with or without extraocular anomalies, 617175 |
| RECQL4 | 100,0% | 100,0% | Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280 |
| RELN | 100,0% | 100,0% | Lissencephaly 2 (Norman-Roberts type), 257320 |
| RERE | 99,9% | 99,9% | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 |
| REV3L | 97,8% | 97,6% | No OMIM Disease ID |
| RFT1 | 100,0% | 100,0% | Congenital disorder of glycosylation, type In, 612015 |
| RFX3 | 100,0% | 100,0% | No OMIM Disease ID |
| RFX4 | 100,0% | 100,0% | No OMIM Disease ID |
| RFX7 | 99,1% | 99,1% | No OMIM Disease ID |
| RHEB | 100,0% | 100,0% | No OMIM Disease ID |
| RHOBTB2 | 100,0% | 100,0% | Developmental and epileptic encephalopathy 64, 618004 |
| RIC1 | 100,0% | 100,0% | CATIFA syndrome, 618761 |
| RIMS2 | 97,8% | 97,8% | Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 |
| RIT1 | 100,0% | 100,0% | Noonan syndrome 8, 615355 |
| RLIM | 100,0% | 100,0% | Tonne-Kalscheuer syndrome, 300978 |
| RMND1 | 100,0% | 100,0% | Combined oxidative phosphorylation deficiency 11, 614922 |
| RMRP | NC | NC | Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250 |
| RNASEH2A | 100,0% | 100,0% | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 91,0% | 91,0% | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 100,0% | 100,0% | Aicardi-Goutieres syndrome 3, 610329 |
| RNASET2 | 100,0% | 100,0% | Leukoencephalopathy, cystic, without megalencephaly, 612951 |
| RNF113A | 100,0% | 100,0% | Trichothiodystrophy 5, nonphotosensitive, 300953 |
| RNF125 | 100,0% | 100,0% | Tenorio syndrome, 616260 |
| RNF13 | 100,0% | 100,0% | Developmental and epileptic encephalopathy 73, 618379 |

| | | | |
|----------|--------|--------|---|
| RNF2 | 100,0% | 100,0% | Luo-Schoch-Yamamoto syndrome, 619460 |
| RNF220 | 100,0% | 100,0% | Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 |
| RNPC3 | 100,0% | 100,0% | Pituitary hormone deficiency, combined or isolated, 7, 618160 |
| RNU4ATAC | NC | NC | Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710 |
| ROGDI | 100,0% | 100,0% | Kohlschutter-Tonz syndrome, 226750 |
| ROR2 | 97,0% | 97,0% | Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310 |
| RORA | 100,0% | 100,0% | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 |
| RPGRI1L | 100,0% | 99,8% | Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113 |
| RPIA | 100,0% | 100,0% | Ribose 5-phosphate isomerase deficiency, 608611 |
| RPL10 | 100,0% | 100,0% | Intellectual developmental disorder, X-linked, syndromic, 35, 300998 |
| RPS19 | 100,0% | 100,0% | Diamond-Blackfan anemia 1, 105650 |
| RPS6KA3 | 100,0% | 99,8% | Intellectual developmental disorder, X-linked 19, 300844 Coffin-Lowry syndrome, 303600 |
| RRM2B | 100,0% | 100,0% | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 |
| RRP7A | 100,0% | 99,9% | ?Microcephaly 28, primary, autosomal recessive, 619453 |
| RSPRY1 | 100,0% | 100,0% | Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 |
| RSRC1 | 100,0% | 100,0% | Intellectual developmental disorder, autosomal recessive 70, 618402 |
| RTEL1 | 100,0% | 100,0% | Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 |
| RTN4IP1 | 100,0% | 100,0% | Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 |
| RTTN | 100,0% | 100,0% | Microcephaly, short stature, and polymicrogyria with seizures, 614833 |
| RUBCN | 100,0% | 100,0% | Spinocerebellar ataxia, autosomal recessive 15, 615705 |
| RUSC2 | 100,0% | 100,0% | Intellectual developmental disorder, autosomal recessive 61, 617773 |
| RXYLT1 | 100,0% | 100,0% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 |
| SALL1 | 100,0% | 100,0% | Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480 |

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

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

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

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

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

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

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

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

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

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

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

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

| | | | |
|--------|--------|--------|---|
| VAR52 | 100,0% | 100,0% | Combined oxidative phosphorylation deficiency 20, 615917 |
| VLDLR | 100,0% | 100,0% | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 |
| VPS11 | 100,0% | 100,0% | ?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683 |
| VPS13B | 99,5% | 99,4% | Cohen syndrome, 216550 |
| VPS16 | 100,0% | 100,0% | Dystonia 30, 619291 |
| VPS35L | 100,0% | 100,0% | Ritscher-Schinzel syndrome 3, 619135 |
| VPS37A | 100,0% | 100,0% | Spastic paraplegia 53, autosomal recessive, 614898 |
| VPS41 | 100,0% | 100,0% | Spinocerebellar ataxia, autosomal recessive 29, 619389 |
| VPS4A | 100,0% | 100,0% | CIMDAG syndrome, 619273 |
| VPS50 | 100,0% | 100,0% | Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 |
| VPS53 | 100,0% | 99,8% | Pontocerebellar hypoplasia, type 2E, 615851 |
| VRK1 | 100,0% | 100,0% | Pontocerebellar hypoplasia type 1A, 607596 |
| VWA3B | 100,0% | 100,0% | ?Spinocerebellar ataxia, autosomal recessive 22, 616948 |
| WAC | 100,0% | 100,0% | Desanto-Shinawi syndrome, 616708 |
| WARS2 | 100,0% | 100,0% | Parkinsonism-dystonia 3, childhood-onset, 619738 Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 |
| WASF1 | 100,0% | 100,0% | Neurodevelopmental disorder with absent language and variable seizures, 618707 |
| WASHC4 | 100,0% | 100,0% | Intellectual developmental disorder, autosomal recessive 43, 615817 |
| WDFY3 | 100,0% | 100,0% | ?Microcephaly 18, primary, autosomal dominant, 617520 |
| WDPCP | 98,1% | 98,1% | ?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 |
| WDR11 | 100,0% | 100,0% | Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 |
| WDR13 | 100,0% | 100,0% | No OMIM Disease ID |
| WDR26 | 97,0% | 94,3% | Skraban-Deardorff syndrome, 617616 |
| WDR37 | 86,5% | 86,5% | Neurooculocardiogenitourinary syndrome, 618652 |
| WDR4 | 100,0% | 100,0% | Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346 |
| WDR45 | 100,0% | 100,0% | Neurodegeneration with brain iron accumulation 5, 300894 |
| WDR45B | 100,0% | 100,0% | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 |
| WDR62 | 100,0% | 100,0% | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 |
| WDR73 | 100,0% | 100,0% | Galloway-Mowat syndrome 1, 251300 |
| WDR81 | 100,0% | 100,0% | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967 |
| WFS1 | 100,0% | 100,0% | Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 |

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

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

Gene symbols used follow 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 is the chemistry used for WES analysis.

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

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

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

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

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

This list is accurate for panel version DG 3.4.0

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