# INTELLECTUAL DISABILITY GENE PANEL DG 3.2.0 ( 1550 genes)

Releasedate: 16-09-2021

| *Gene* | *Agilent V5 covered >10x* | *Agilent V5 covered >20x* | *TWIST covered >10x* | *TWIST covered >20x* | *Associated Phenotype Description and OMIM disease ID* |
|---|---|---|---|---|---|
| AAAS | 100 | 99,4 | 100 | 100 | Achalasia-addisonianism-alacrimia syndrome, 231550 |
| AARS1 | 100 | 99,7 | 100 | 99,9 | Developmental and epileptic encephalopathy 29, 616339<br>Charcot-Marie-Tooth disease, axonal, type 2N, 613287 |
| AASS | 99,9 | 99,8 | 100 | 99,9 | Hyperlysinemia, 238700 |
| ABAT | 99,9 | 97,8 | 100 | 100 | GABA-transaminase deficiency, 613163 |
| ABCA2 | 99,9 | 99,2 | 100 | 99,9 | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 |
| ABCC8 | 100 | 99,5 | 100 | 100 | Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857<br>Diabetes mellitus, transient neonatal 2, 610374<br>Diabetes mellitus, noninsulin-dependent, 125853<br>Hypoglycemia of infancy, leucine-sensitive, 240800<br>Hyperinsulinemic hypoglycemia, familial, 1, 256450 |
| ABCC9 | 100 | 99,8 | 100 | 100 | Cardiomyopathy, dilated, 1O, 608569<br>Hypertrichotic osteochondrodysplasia, 239850<br>?Atrial fibrillation, familial, 12, 614050 |
| ABCD1 | 76 | 72,6 | 100 | 100 | Adrenoleukodystrophy, 300100<br>Adrenomyeloneuropathy, adult, 300100 |
| ABCD4 | 99,8 | 97,7 | 100 | 100 | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| ABHD5 | 100 | 100 | 100 | 100 | Chanarin-Dorfman syndrome, 275630 |
| ACAD9 | 100 | 99,8 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACADS | 100 | 99,4 | 100 | 100 | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 |
| ACADSB | 99,8 | 97,9 | 100 | 100 | 2-methylbutyrylglycinuria, 610006 |
| ACAT1 | 99,6 | 97,9 | 100 | 99,7 | Alpha-methylacetoacetic aciduria, 203750 |
| ACO2 | 94,1 | 86,3 | 100 | 100 | ?Optic atrophy 9, 616289<br>Infantile cerebellar-retinal degeneration, 614559 |
| ACOX1 | 100 | 99,3 | 100 | 100 | Mitchell syndrome, 618960<br>Peroxisomal acyl-CoA oxidase deficiency, 264470 |

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| ACSF3 | 100 | 99,5 | 100 | 100 | Combined malonic and methylmalonic aciduria, 614265 |
| ACSL4 | 98,3 | 94,2 | 100 | 99,6 | Intellectual developmental disorder, X-linked 63, 300387 |
| ACTB | 99,9 | 97,2 | 100 | 100 | Baraitser-Winter syndrome 1, 243310<br>?Dystonia, juvenile-onset, 607371 |
| ACTG1 | 100 | 100 | 100 | 100 | Deafness, autosomal dominant 20/26, 604717<br>Baraitser-Winter syndrome 2, 614583 |
| ACTL6A | 99,8 | 98,9 | 100 | 99,9 | No OMIM disease ID |
| ACTL6B | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 76, 618468<br>Intellectual developmental disorder with severe speech and ambulation defects, 618470 |
| ACVR1 | 100 | 99,9 | 100 | 100 | Fibrodysplasia ossificans progressiva, 135100 |
| ACY1 | 100 | 99,7 | 100 | 100 | Aminoacylase 1 deficiency, 609924 |
| ADAM22 | 99,8 | 99,6 | 100 | 100 | Developmental and epileptic encephalopathy 61, 617933 |
| ADAR | 100 | 99,4 | 100 | 100 | Dyschromatosis symmetrica hereditaria, 127400<br>Aicardi-Goutieres syndrome 6, 615010 |
| ADARB1 | 97,2 | 95,3 | 95,1 | 95,1 | Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 |
| ADAT3 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 |
| ADD3 | 99,8 | 98,8 | 100 | 100 | Cerebral palsy, spastic quadriplegic, 3, 617008 |
| ADGRG1 | 100 | 100 | 100 | 100 | Polymicrogyria, bilateral frontoparietal, 606854<br>Polymicrogyria, bilateral perisylvian, 615752 |
| ADK | 83,3 | 79,7 | 84,5 | 84,5 | Hypermethioninemia due to adenosine kinase deficiency, 614300 |
| ADNP | 90,5 | 90,5 | 95,4 | 95,4 | Helsmoortel-van der Aa syndrome, 615873 |
| ADPRS | 100 | 99,9 | 100 | 100 | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 |
| ADSL | 99,2 | 98,6 | 100 | 100 | Adenylosuccinase deficiency, 103050 |
| AFF2 | 99,8 | 98,8 | 100 | 99,6 | Intellectual developmental disorder, X-linked 109, 309548 |
| AFF3 | 98,6 | 97,9 | 100 | 100 | KINSSHIP syndrome, 619297 |
| AFF4 | 99,8 | 98,2 | 100 | 100 | CHOPS syndrome, 616368 |
| AFG3L2 | 94,6 | 86,3 | 100 | 100 | Spastic ataxia 5, autosomal recessive, 614487<br>Optic atrophy 12, 618977<br>Spinocerebellar ataxia 28, 610246 |
| AGA | 100 | 99,9 | 100 | 100 | Aspartylglucosaminuria, 208400 |
| AGAP1 | 97,1 | 91 | 100 | 99,5 | No OMIM disease ID |
| AGMO | 99 | 92,4 | 100 | 99,9 | No OMIM disease ID |

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| AGO2 | 99,1 | 99,1 | 99,7 | 99,3 | Lessel-Kreienkamp syndrome, 619149 |
| AGTPBP1 | 96,3 | 94,2 | 100 | 100 | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 |
| AHCY | 99,9 | 98,8 | 100 | 100 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 |
| AHDC1 | 100 | 98,9 | 100 | 100 | Xia-Gibbs syndrome, 615829 |
| AHI1 | 99,4 | 97,4 | 100 | 100 | Joubert syndrome 3, 608629 |
| AHSG | 100 | 99,8 | 100 | 100 | ?Alopecia-mental retardation syndrome 1, 203650 |
| AIFM1 | 99,9 | 97,8 | 100 | 100 | Combined oxidative phosphorylation deficiency 6, 300816<br>Cowchock syndrome, 310490<br>Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232<br>Deafness, X-linked 5, 300614 |
| AIMP1 | 99,2 | 92,5 | 100 | 99,9 | Leukodystrophy, hypomyelinating, 3, 260600 |
| AIMP2 | 89,4 | 86 | 100 | 99,9 | Leukodystrophy, hypomyelinating, 17, 618006 |
| AKT3 | 97,4 | 92,4 | 100 | 100 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 |
| ALDH18A1 | 100 | 99,9 | 100 | 100 | Spastic paraplegia 9A, autosomal dominant, 601162<br>Cutis laxa, autosomal recessive, type IIIA, 219150<br>Spastic paraplegia 9B, autosomal recessive, 616586<br>Cutis laxa, autosomal dominant 3, 616603 |
| ALDH3A2 | 88,8 | 88,4 | 93,2 | 93,2 | Sjogren-Larsson syndrome, 270200 |
| ALDH4A1 | 100 | 99,7 | 100 | 100 | Hyperprolinemia, type II, 239510 |
| ALDH5A1 | 92,4 | 83,5 | 100 | 100 | Succinic semialdehyde dehydrogenase deficiency, 271980 |
| ALDH7A1 | 91,1 | 84,5 | 100 | 100 | Epilepsy, pyridoxine-dependent, 266100 |
| ALG1 | 53,6 | 46,9 | 100 | 100 | Congenital disorder of glycosylation, type Ik, 608540 |
| ALG11 | 96,8 | 96,8 | 96,8 | 96,8 | Congenital disorder of glycosylation, type Ip, 613661 |
| ALG12 | 100 | 99,9 | 100 | 100 | Congenital disorder of glycosylation, type Ig, 607143 |
| ALG13 | 97,4 | 90 | 99,9 | 99,4 | ?Congenital disorder of glycosylation, type Is, 300884<br>Developmental and epileptic encephalopathy 36, 300884 |
| ALG14 | 100 | 99,9 | 100 | 100 | Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031<br>Myopathy, epilepsy, and progressive cerebral atrophy, 619036<br>?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 |
| ALG2 | 100 | 100 | 100 | 100 | ?Congenital disorder of glycosylation, type Ii, 607906<br>Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 |
| ALG3 | 100 | 99,5 | 100 | 100 | Congenital disorder of glycosylation, type Id, 601110 |

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| ALG6 | 98,2 | 93,7 | 100 | 99,9 | Congenital disorder of glycosylation, type Ic, 603147 |
| ALG8 | 96,6 | 95,9 | 96,6 | 96,6 | Congenital disorder of glycosylation, type Ih, 608104<br>Polycystic liver disease 3 with or without kidney cysts, 617874 |
| ALG9 | 99,9 | 99,3 | 100 | 100 | Gillessen-Kaesbach-Nishimura syndrome, 263210<br>Congenital disorder of glycosylation, type Il, 608776 |
| ALKBH8 | 99,7 | 98,6 | 100 | 100 | Intellectual developmental disorder, autosomal recessive 71, 618504 |
| ALMS1 | 99,7 | 99,5 | 100 | 100 | Alstrom syndrome, 203800 |
| ALX3 | 80,2 | 72,8 | 100 | 100 | Frontonasal dysplasia 1, 136760 |
| ALX4 | 100 | 99,9 | 100 | 100 | Parietal foramina 2, 609597<br>Frontonasal dysplasia 2, 613451 |
| AMER1 | 99,6 | 96,6 | 100 | 100 | Osteopathia striata with cranial sclerosis, 300373 |
| AMMECR1 | 99,9 | 98,4 | 100 | 100 | Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 |
| AMPD2 | 99,8 | 99 | 100 | 100 | ?Spastic paraplegia 63, 615686<br>Pontocerebellar hypoplasia, type 9, 615809 |
| AMT | 100 | 100 | 100 | 100 | Glycine encephalopathy, 605899 |
| ANK3 | 99,3 | 99 | 100 | 100 | Mental retardation, autosomal recessive, 37, 615493 |
| ANKH | 100 | 99,9 | 100 | 100 | Chondrocalcinosis 2, 118600<br>Craniometaphyseal dysplasia, 123000 |
| ANKLE2 | 100 | 99,3 | 100 | 99,7 | Microcephaly 16, primary, autosomal recessive, 616681 |
| ANKRD11 | 97 | 94 | 100 | 100 | KBG syndrome, 148050 |
| ANKRD17 | 99,4 | 98,2 | 100 | 100 | Chopra-Amiel-Gordon syndrome, 619504 |
| ANKS1B | 99,9 | 99,3 | 100 | 100 | No OMIM disease ID |
| ANO10 | 99,2 | 96,6 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 10, 613728 |
| ANTXR1 | 99,9 | 99,1 | 100 | 100 | GAPO syndrome, 230740 |
| AP1S1 | 99,9 | 99,4 | 100 | 100 | MEDNIK syndrome, 609313 |
| AP1S2 | 73,8 | 66,8 | 100 | 98,8 | Pettigrew syndrome, 304340 |
| AP2M1 | 100 | 99,9 | 100 | 100 | Intellectual developmental disorder 60 with seizures, 618587 |
| AP2S1 | 90,4 | 90 | 100 | 100 | Hypocalciuric hypercalcemia, type III, 600740 |
| AP3B1 | 99,2 | 96,4 | 100 | 99,9 | Hermansky-Pudlak syndrome 2, 608233 |
| AP3B2 | 93,3 | 89,8 | 99,9 | 99 | Developmental and epileptic encephalopathy 48, 617276 |
| AP3D1 | 99,6 | 98,4 | 100 | 100 | ?Hermansky-Pudlak syndrome 10, 617050 |

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| AP4B1 | 99,8 | 98,5 | 100 | 100 | Spastic paraplegia 47, autosomal recessive, 614066 |
| AP4E1 | 99,5 | 98,6 | 100 | 99,9 | Stuttering, familial persistent, 1, 184450<br>Spastic paraplegia 51, autosomal recessive, 613744 |
| AP4M1 | 99,8 | 97,8 | 100 | 100 | Spastic paraplegia 50, autosomal recessive, 612936 |
| AP4S1 | 78,9 | 71 | 87,9 | 87,9 | Spastic paraplegia 52, autosomal recessive, 614067 |
| APC2 | 98,3 | 94,8 | 99,7 | 98,5 | Cortical dysplasia, complex, with other brain malformations 10, 618677<br>?Sotos syndrome 3, 617169 |
| APTX | 94,1 | 90,6 | 100 | 100 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 |
| ARCN1 | 96,8 | 96,6 | 96,7 | 96,6 | Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164 |
| ARF1 | 100 | 99,9 | 100 | 100 | Periventricular nodular heterotopia 8, 618185 |
| ARFGEF2 | 99,7 | 98,7 | 100 | 100 | Periventricular heterotopia with microcephaly, 608097 |
| ARG1 | 92,9 | 92,9 | 92,9 | 92,7 | Argininemia, 207800 |
| ARHGAP31 | 99,7 | 98,2 | 100 | 100 | Adams-Oliver syndrome 1, 100300 |
| ARHGAP35 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| ARHGEF6 | 99 | 94,8 | 100 | 99,8 | No OMIM disease ID |
| ARHGEF9 | 76,3 | 72,5 | 97,2 | 97,1 | Developmental and epileptic encephalopathy 8, 300607 |
| ARID1A | 98,3 | 96 | 100 | 100 | Coffin-Siris syndrome 2, 614607 |
| ARID1B | 96,2 | 94,6 | 97,8 | 96,9 | Coffin-Siris syndrome 1, 135900 |
| ARID2 | 99,7 | 98,2 | 100 | 100 | Coffin-Siris syndrome 6, 617808 |
| ARL13B | 100 | 99,3 | 100 | 100 | Joubert syndrome 8, 612291 |
| ARL6 | 99,1 | 98,4 | 100 | 100 | Retinitis pigmentosa 55, 613575<br>Bardet-Biedl syndrome 3, 600151 |
| ARMC9 | 99,9 | 99,4 | 100 | 100 | Joubert syndrome 30, 617622 |
| ARSA | 100 | 99,8 | 100 | 100 | Metachromatic leukodystrophy, 250100 |
| ARSL | 98,9 | 92,4 | 100 | 99,8 | Chondrodysplasia punctata, X-linked recessive, 302950 |
| ARV1 | 99,9 | 98,8 | 100 | 99,9 | Developmental and epileptic encephalopathy 38, 617020 |
| ARX | 82,1 | 67,5 | 91,4 | 86,6 | Proud syndrome, 300004<br>Hydranencephaly with abnormal genitalia, 300215<br>Partington syndrome, 309510<br>Developmental and epileptic encephalopathy 1, 308350<br>Lissencephaly, X-linked 2, 300215<br>Intellectual developmental disorder, X-linked 29, 300419 |

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| ASAH1 | 99,1 | 97,3 | 100 | 100 | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950<br>Farber lipogranulomatosis, 228000 |
| ASH1L | 98,7 | 98,6 | 98,7 | 98,7 | Mental retardation, autosomal dominant 52, 617796 |
| ASL | 100 | 99,7 | 100 | 100 | Argininosuccinic aciduria, 207900 |
| ASNS | 98,1 | 91,2 | 100 | 100 | Asparagine synthetase deficiency, 615574 |
| ASPA | 99,9 | 99,1 | 100 | 100 | Canavan disease, 271900 |
| ASPM | 99,4 | 97,9 | 100 | 99,9 | Microcephaly 5, primary, autosomal recessive, 608716 |
| ASS1 | 93,2 | 83,2 | 100 | 100 | Citrullinemia, 215700 |
| ASXL1 | 99,8 | 98,9 | 100 | 100 | Myelodysplastic syndrome, somatic, 614286<br>Bohring-Opitz syndrome, 605039 |
| ASXL2 | 99,9 | 99,5 | 100 | 100 | Shashi-Pena syndrome, 617190 |
| ASXL3 | 99,9 | 99,6 | 100 | 100 | Bainbridge-Ropers syndrome, 615485 |
| ATAD1 | 99,1 | 91,6 | 100 | 100 | Hyperekplexia 4, 618011 |
| ATAD3A | 91,4 | 86,7 | 100 | 100 | Harel-Yoon syndrome, 617183<br>Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 |
| ATIC | 99,8 | 99,1 | 100 | 100 | AICA-ribosiduria due to ATIC deficiency, 608688 |
| ATL1 | 99,9 | 99,5 | 100 | 99,8 | Spastic paraplegia 3A, autosomal dominant, 182600<br>Neuropathy, hereditary sensory, type ID, 613708 |
| ATN1 | 99,8 | 98 | 100 | 100 | Dentatorubral-pallidoluysian atrophy, 125370<br>Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 |
| ATP13A2 | 100 | 99,6 | 100 | 100 | Spastic paraplegia 78, autosomal recessive, 617225<br>Kufor-Rakeb syndrome, 606693 |
| ATP1A1 | 100 | 99,8 | 100 | 100 | Hypomagnesemia, seizures, and mental retardation 2, 618314<br>Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 |
| ATP1A2 | 100 | 99,8 | 100 | 100 | Alternating hemiplegia of childhood 1, 104290<br>Migraine, familial basilar, 602481<br>Migraine, familial hemiplegic, 2, 602481 |
| ATP1A3 | 100 | 99,9 | 100 | 100 | Alternating hemiplegia of childhood 2, 614820<br>Dystonia-12, 128235<br>CAPOS syndrome, 601338 |
| ATP2A2 | 100 | 99,6 | 100 | 100 | Acrokeratosis verruciformis, 101900<br>Darier disease, 124200 |
| ATP6AP1 | 98,2 | 93 | 100 | 100 | Immunodeficiency 47, 300972 |

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| ATP6AP2 | 89,9 | 69,7 | 100 | 99,8 | Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423<br>?Parkinsonism with spasticity, X-linked, 300911<br>Congenital disorder of glycosylation, type IIr, 301045 |
| ATP6V0A1 | 99,8 | 98,6 | 100 | 100 | No OMIM disease ID |
| ATP6V0A2 | 99,9 | 98,7 | 100 | 100 | Wrinkly skin syndrome, 278250<br>Cutis laxa, autosomal recessive, type IIA, 219200 |
| ATP6V0C | 100 | 100 | 100 | 100 | No OMIM disease ID |
| ATP6V1A | 99,8 | 98,4 | 100 | 100 | Cutis laxa, autosomal recessive, type IID, 617403<br>Developmental and epileptic encephalopathy 93, 618012 |
| ATP6V1B2 | 99,9 | 99,3 | 100 | 100 | Zimmermann-Laband syndrome 2, 616455<br>Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 |
| ATP7A | 98,7 | 96 | 100 | 99,9 | Occipital horn syndrome, 304150<br>Spinal muscular atrophy, distal, X-linked 3, 300489<br>Menkes disease, 309400 |
| ATP8A2 | 100 | 99,6 | 100 | 100 | ?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 |
| ATR | 99,7 | 99 | 100 | 100 | Seckel syndrome 1, 210600<br>?Cutaneous telangiectasia and cancer syndrome, familial, 614564 |
| ATRX | 98,7 | 95,2 | 100 | 99,9 | Alpha-thalassemia/mental retardation syndrome, 301040<br>Alpha-thalassemia myelodysplasia syndrome, somatic, 300448<br>Mental retardation-hypotonic facies syndrome, X-linked, 309580 |
| ATXN2L | 98,3 | 94,7 | 100 | 100 | No OMIM disease ID |
| AUH | 99,7 | 99,4 | 100 | 99,9 | 3-methylglutaconic aciduria, type I, 250950 |
| AUTS2 | 98,8 | 96,9 | 100 | 100 | Mental retardation, autosomal dominant 26, 615834 |
| AVPR2 | 100 | 99,8 | 100 | 100 | Diabetes insipidus, nephrogenic, 1, 304800<br>Nephrogenic syndrome of inappropriate antidiuresis, 300539 |
| B3GALNT2 | 94,3 | 89,8 | 92,5 | 92,5 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 |
| B3GALT6 | 77 | 73 | 91,7 | 81 | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349<br>Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640<br>Al-Gazali syndrome, 609465 |
| B3GLCT | 99,7 | 98,2 | 100 | 99,4 | Peters-plus syndrome, 261540 |
| B4GALNT1 | 98,3 | 93,5 | 100 | 100 | Spastic paraplegia 26, autosomal recessive, 609195 |
| B4GALT1 | 100 | 99,3 | 100 | 100 | Congenital disorder of glycosylation, type IId, 607091 |
| B4GALT7 | 99,7 | 96,8 | 100 | 99,4 | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 |
| B4GAT1 | 100 | 100 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 |

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| B9D1 | 85,2 | 85,2 | 95,8 | 94 | ?Meckel syndrome 9, 614209<br>Joubert syndrome 27, 617120 |
| B9D2 | 100 | 100 | 100 | 100 | ?Meckel syndrome 10, 614175<br>Joubert syndrome 34, 614175 |
| BAZ2B | 99,6 | 98,8 | 100 | 99,9 | No OMIM disease ID |
| BBS1 | 100 | 100 | 100 | 100 | Bardet-Biedl syndrome 1, 209900 |
| BBS10 | 100 | 99,9 | 100 | 100 | Bardet-Biedl syndrome 10, 615987 |
| BBS12 | 100 | 100 | 100 | 100 | Bardet-Biedl syndrome 12, 615989 |
| BBS2 | 99,4 | 98 | 100 | 100 | Retinitis pigmentosa 74, 616562<br>Bardet-Biedl syndrome 2, 615981 |
| BBS4 | 99,9 | 98,9 | 100 | 99,9 | Bardet-Biedl syndrome 4, 615982 |
| BBS5 | 98,4 | 94,7 | 100 | 100 | Bardet-Biedl syndrome 5, 615983 |
| BBS7 | 99 | 96,5 | 100 | 99,9 | Bardet-Biedl syndrome 7, 615984 |
| BBS9 | 92 | 89 | 95,8 | 95,8 | Bardet-Biedl syndrome 9, 615986 |
| BCAP31 | 92,1 | 79,1 | 100 | 99,6 | Deafness, dystonia, and cerebral hypomyelination, 300475 |
| BCAS3 | 99,1 | 98,9 | 100 | 100 | No OMIM disease ID |
| BCKDHA | 99,8 | 97,9 | 100 | 100 | Maple syrup urine disease, type Ia, 248600 |
| BCKDHB | 99,8 | 95,4 | 100 | 100 | Maple syrup urine disease, type Ib, 248600 |
| BCKDK | 100 | 100 | 100 | 100 | Branched-chain ketoacid dehydrogenase kinase deficiency, 614923 |
| BCL11A | 97,3 | 96 | 100 | 100 | Dias-Logan syndrome, 617101 |
| BCL11B | 99,6 | 96,5 | 99,4 | 97,9 | Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092<br>Immunodeficiency 49, 617237 |
| BCOR | 99,2 | 95,8 | 100 | 100 | Microphthalmia, syndromic 2, 300166 |
| BCORL1 | 99,4 | 97,7 | 100 | 100 | Shukla-Vernon syndrome, 301029 |
| BCS1L | 100 | 100 | 100 | 100 | GRACILE syndrome, 603358<br>Mitochondrial complex III deficiency, nuclear type 1, 124000<br>Bjornstad syndrome, 262000 |
| BICRA | 99,8 | 98,4 | 100 | 100 | Coffin-Siris syndrome 12, 619325 |
| BLM | 99,3 | 97,7 | 100 | 100 | Bloom syndrome, 210900 |
| BLOC1S1 | 100 | 99,1 | 100 | 100 | No OMIM disease ID |
| BOLA3 | 99 | 86,7 | 100 | 100 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 |

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| BPTF | 96,1 | 94,3 | 99,6 | 98,4 | Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 |
| BRAF | 89,4 | 77,6 | 100 | 100 | Melanoma, malignant, somatic, 155600<br>LEOPARD syndrome 3, 613707<br>Cardiofaciocutaneous syndrome, 115150<br>Adenocarcinoma of lung, somatic, 211980<br>Noonan syndrome 7, 613706<br>Colorectal cancer, somatic, 114500<br>Nonsmall cell lung cancer, somatic, 211980 |
| BRAT1 | 99,9 | 98,9 | 100 | 100 | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056<br>Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 |
| BRF1 | 99,8 | 98,4 | 100 | 100 | Cerebellofaciodental syndrome, 616202 |
| BRPF1 | 100 | 100 | 100 | 100 | Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 |
| BRSK2 | 99,4 | 96,9 | 100 | 100 | No OMIM disease ID |
| BRWD3 | 98,7 | 95,3 | 100 | 99,8 | Intellectual developmental disorder, X-linked 93, 300659 |
| BSCL2 | 100 | 99,9 | 100 | 100 | Lipodystrophy, congenital generalized, type 2, 269700<br>Neuropathy, distal hereditary motor, type VC, 619112<br>Silver spastic paraplegia syndrome, 270685<br>Encephalopathy, progressive, with or without lipodystrophy, 615924 |
| BTD | 83 | 82,9 | 83,1 | 83,1 | Biotinidase deficiency, 253260 |
| BUB1B | 99,3 | 98,3 | 100 | 100 | Colorectal cancer, somatic, 114500<br>Mosaic variegated aneuploidy syndrome 1, 257300 |
| C12orf4 | 99,6 | 99,5 | 100 | 100 | Mental retardation, autosomal recessive 66, 618221 |
| C12orf57 | 100 | 98,6 | 100 | 100 | Temtamy syndrome, 218340 |
| C2CD3 | 95,8 | 95,4 | 95,9 | 95,9 | Orofaciodigital syndrome XIV, 615948 |
| CA2 | 100 | 100 | 100 | 100 | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 |
| CA5A | 87,6 | 85,6 | 87,7 | 87,7 | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 |
| CA8 | 99,4 | 96,6 | 100 | 100 | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 |
| CACNA1A | 93,1 | 88,4 | 100 | 99,9 | Developemental and epileptic encephalopathy 42, 617106<br>Spinocerebellar ataxia 6, 183086<br>Episodic ataxia, type 2, 108500<br>Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500<br>Migraine, familial hemiplegic, 1, 141500 |
| CACNA1B | 98 | 96,1 | 99,3 | 98,2 | Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 |

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| CACNA1C | 99,9 | 99,2 | 100 | 100 | Timothy syndrome, 601005<br>Long QT syndrome 8, 618447<br>Brugada syndrome 3, 611875 |
| CACNA1D | 97,9 | 97,7 | 100 | 100 | Primary aldosteronism, seizures, and neurologic abnormalities, 615474<br>Sinoatrial node dysfunction and deafness, 614896 |
| CACNA1E | 100 | 99,8 | 100 | 100 | Developmental and epileptic encephalopathy 69, 618285 |
| CACNA1G | 100 | 99,2 | 100 | 100 | Spinocerebellar ataxia 42, 616795<br>Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 |
| CACNA2D2 | 93,8 | 93,1 | 99,6 | 98,4 | Cerebellar atrophy with seizures and variable developmental delay, 618501 |
| CAD | 99,8 | 98,5 | 100 | 100 | Developmental and epileptic encephalopathy 50, 616457 |
| CAMK2A | 99,8 | 98,4 | 99,9 | 99,6 | Mental retardation, autosomal dominant 53, 617798<br>?Mental retardation, autosomal recessive 63, 618095 |
| CAMK2B | 100 | 99,4 | 100 | 99,7 | Mental retardation, autosomal dominant 54, 617799 |
| CAMK2G | 99,9 | 98,6 | 100 | 100 | Mental retardation, autosomal dominant 59, 618522 |
| CAMTA1 | 99,6 | 99 | 100 | 100 | Cerebellar ataxia, nonprogressive, with mental retardation, 614756 |
| CANT1 | 100 | 100 | 100 | 100 | Desbuquois dysplasia 1, 251450<br>Epiphyseal dysplasia, multiple, 7, 617719 |
| CAPN15 | 99,8 | 98,1 | 100 | 100 | Oculogastrointestinal neurodevelopmental syndrome, 619318 |
| CARS2 | 100 | 100 | 100 | 99,5 | Combined oxidative phosphorylation deficiency 27, 616672 |
| CASK | 97,2 | 93,9 | 100 | 99,9 | Mental retardation, with or without nystagmus, 300422<br>Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749<br>FG syndrome 4, 300422 |
| CBL | 97,3 | 96,9 | 100 | 100 | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563<br>?Juvenile myelomonocytic leukemia, 607785 |
| CBS | 99,9 | 98,5 | 100 | 100 | Thrombosis, hyperhomocysteinemic, 236200<br>Homocystinuria, B6-responsive and nonresponsive types, 236200 |
| CC2D1A | 100 | 99,6 | 100 | 100 | Mental retardation, autosomal recessive 3, 608443 |
| CC2D2A | 98,3 | 96,6 | 97,1 | 97 | COACH syndrome 2, 619111<br>Meckel syndrome 6, 612284<br>Joubert syndrome 9, 612285 |
| CCBE1 | 99,9 | 98,8 | 100 | 100 | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 |
| CCDC115 | 95,8 | 90,2 | 100 | 100 | Congenital disorder of glycosylation, type IIo, 616828 |
| CCDC174 | 99,5 | 96,7 | 100 | 100 | Hypotonia, infantile, with psychomotor retardation, 616816 |
| CCDC186 | 99 | 95,6 | 100 | 99,8 | No OMIM disease ID |

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|---|---|---|---|---|---|
| CCDC22 | 99 | 95,4 | 100 | 100 | Ritscher-Schinzel syndrome 2, 300963 |
| CCDC32 | 99,9 | 98,5 | 100 | 100 | Cardiofacioneurodevelopmental syndrome, 619123 |
| CCDC47 | 99,9 | 97,4 | 100 | 100 | Trichohepatoneurodevelopmental syndrome, 618268 |
| CCDC88A | 95,9 | 91,8 | 97,5 | 97,3 | ?PEHO syndrome-like, 617507 |
| CCDC88C | 99,9 | 99,3 | 100 | 100 | ?Spinocerebellar ataxia 40, 616053<br>Hydrocephalus, congenital, 1, 236600 |
| CCND2 | 100 | 100 | 100 | 100 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 |
| CCNK | 92,4 | 89 | 100 | 99,2 | ?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 |
| CDC42 | 96,3 | 87,9 | 100 | 100 | Takenouchi-Kosaki syndrome, 616737 |
| CDC42BPB | 99,9 | 98,5 | 100 | 100 | No OMIM disease ID |
| CDC6 | 100 | 99,9 | 100 | 100 | ?Meier-Gorlin syndrome 5, 613805 |
| CDH11 | 100 | 100 | 100 | 100 | Elsahy-Waters syndrome, 211380 |
| CDH15 | 100 | 99,2 | 100 | 100 | Mental retardation, autosomal dominant 3, 612580 |
| CDK10 | 100 | 99,5 | 100 | 100 | Al Kaissi syndrome, 617694 |
| CDK13 | 97,7 | 91,7 | 100 | 99,9 | Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 |
| CDK19 | 99,8 | 99,3 | 100 | 100 | Developmental and epileptic encephalopathy 87, 618916 |
| CDK5RAP2 | 99,6 | 98,5 | 100 | 100 | Microcephaly 3, primary, autosomal recessive, 604804 |
| CDK8 | 99,6 | 96,7 | 100 | 100 | Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 |
| CDKL5 | 91,4 | 89 | 92,2 | 91,1 | Developmental and epileptic encephalopathy 2, 300672 |
| CDKN1C | 89,9 | 81,6 | 98,9 | 95,8 | IMAGE syndrome, 614732<br>Beckwith-Wiedemann syndrome, 130650 |
| CDON | 99,9 | 98,6 | 100 | 99,9 | Holoprosencephaly 11, 614226 |
| CELF2 | 94,9 | 94,4 | 100 | 100 | No OMIM disease ID |
| CENPF | 99,4 | 96,9 | 100 | 100 | Stromme syndrome, 243605 |
| CENPJ | 99,8 | 98,7 | 100 | 100 | Microcephaly 6, primary, autosomal recessive, 608393<br>?Seckel syndrome 4, 613676 |
| CEP104 | 99,9 | 98 | 100 | 100 | Joubert syndrome 25, 616781 |
| CEP120 | 99,9 | 99,6 | 100 | 100 | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300<br>Joubert syndrome 31, 617761 |
| CEP135 | 98,3 | 90,1 | 100 | 99,9 | Microcephaly 8, primary, autosomal recessive, 614673 |

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|---|---|---|---|---|---|
| CEP152 | 99,5 | 98 | 100 | 100 | Microcephaly 9, primary, autosomal recessive, 614852<br>Seckel syndrome 5, 613823 |
| CEP290 | 96,2 | 90,8 | 100 | 99,9 | Leber congenital amaurosis 10, 611755<br>Joubert syndrome 5, 610188<br>Senior-Loken syndrome 6, 610189<br>?Bardet-Biedl syndrome 14, 615991<br>Meckel syndrome 4, 611134 |
| CEP41 | 98,8 | 93,4 | 100 | 100 | Joubert syndrome 15, 614464 |
| CEP55 | 100 | 99,8 | 100 | 100 | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 |
| CEP57 | 97,6 | 89,3 | 100 | 100 | Mosaic variegated aneuploidy syndrome 2, 614114 |
| CEP83 | 99 | 96,6 | 100 | 99,9 | Nephronophthisis 18, 615862 |
| CEP85L | 99 | 96,5 | 100 | 99,9 | Lissencephaly 10, 618873 |
| CEP89 | 95,8 | 94,5 | 100 | 100 | No OMIM disease ID |
| CERT1 | 89,8 | 86,5 | 100 | 100 | Mental retardation, autosomal dominant 34, 616351 |
| CHAMP1 | 100 | 100 | 100 | 100 | Mental retardation, autosomal dominant 40, 616579 |
| CHD1 | 98,6 | 93 | 100 | 99,9 | Pilarowski-Bjornsson syndrome, 617682 |
| CHD2 | 99,3 | 98,6 | 100 | 100 | Develomental and epileptic encephalopathy 94, 615369 |
| CHD3 | 96,3 | 92 | 99,7 | 99,4 | Snijders Blok-Campeau syndrome, 618205 |
| CHD4 | 100 | 99,6 | 100 | 100 | Sifrim-Hitz-Weiss syndrome, 617159 |
| CHD5 | 99,6 | 96,1 | 100 | 99,9 | No OMIM disease ID |
| CHD7 | 100 | 99,2 | 100 | 100 | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370<br>CHARGE syndrome, 214800 |
| CHD8 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| CHKB | 100 | 99,6 | 100 | 100 | Muscular dystrophy, congenital, megaconial type, 602541 |
| CHMP1A | 100 | 99,6 | 100 | 100 | Pontocerebellar hypoplasia, type 8, 614961 |
| CHRNA4 | 99 | 96,5 | 100 | 100 | Epilepsy, nocturnal frontal lobe, 1, 600513 |
| CIC | 63,4 | 63,3 | 100 | 100 | Mental retardation, autosomal dominant 45, 617600 |
| CIT | 99,8 | 98,2 | 100 | 100 | Microcephaly 17, primary, autosomal recessive, 617090 |
| CKAP2L | 99,5 | 98,3 | 100 | 100 | Filippi syndrome, 272440 |
| CLCN3 | 98 | 94,2 | 96,7 | 96,7 | Neurodevelopmental disorder with seizures and brain abnormalities, 619517<br>Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 |

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|---|---|---|---|---|---|
| CLCN4 | 99,9 | 97,7 | 100 | 100 | Raynaud-Claes syndrome, 300114 |
| CLDN11 | 100 | 99,9 | 100 | 100 | Leukodystrophy, hypomyelinating, 22, 619328 |
| CLIC2 | 99,3 | 94,9 | 100 | 100 | ?Intellectual developmental disorder, X-linked syndromic 32, 300886 |
| CLIP1 | 99,8 | 98,7 | 100 | 100 | No OMIM disease ID |
| CLN3 | 92,5 | 92,4 | 92,5 | 92,5 | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| CLN5 | 69 | 66,3 | 71,8 | 71,6 | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 99,9 | 98,9 | 100 | 100 | Ceroid lipofuscinosis, neuronal, 6, 601780<br>Ceroid lipofuscinosis, neuronal, 4A (Kufs type), autosomal recessive, 204300 |
| CLN8 | 83,5 | 83,5 | 100 | 100 | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003<br>Ceroid lipofuscinosis, neuronal, 8, 600143 |
| CLP1 | 100 | 100 | 100 | 100 | Pontocerebellar hypoplasia, type 10, 615803 |
| CLPB | 94,9 | 94 | 100 | 100 | 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 |
| CLTC | 99,9 | 99,7 | 100 | 100 | Mental retardation, autosomal dominant 56, 617854 |
| CNKSR2 | 95,1 | 89 | 100 | 99,8 | Intellectual developmental disorder, X-linked, syndromic, Houge type, 301008 |
| CNNM2 | 100 | 99,9 | 100 | 100 | Hypomagnesemia 6, renal, 613882<br>Hypomagnesemia, seizures, and mental retardation, 616418 |
| CNOT1 | 100 | 99,8 | 100 | 100 | Vissers-Bodmer syndrome, 619033<br>Holoprosencephaly 12, with or without pancreatic agenesis, 618500 |
| CNOT2 | 99,9 | 99,6 | 100 | 99,9 | Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 |
| CNOT3 | 100 | 100 | 100 | 100 | Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 |
| CNPY3 | 100 | 99,7 | 100 | 100 | Developmental and epileptic encephalopathy 60, 617929 |
| CNTNAP1 | 100 | 99,8 | 100 | 100 | Lethal congenital contracture syndrome 7, 616286<br>Hypomyelinating neuropathy, congenital, 3, 618186 |
| CNTNAP2 | 100 | 99,5 | 100 | 100 | Pitt-Hopkins like syndrome 1, 610042<br>Cortical dysplasia-focal epilepsy syndrome, 610042 |
| COA8 | 81,9 | 80,8 | 93,7 | 93,4 | Mitochondrial complex IV deficiency, nuclear type 17, 619061 |
| COASY | 100 | 100 | 100 | 100 | Pontocerebellar hypoplasia, type 12, 618266<br>Neurodegeneration with brain iron accumulation 6, 615643 |
| COG1 | 100 | 99,9 | 100 | 100 | Congenital disorder of glycosylation, type IIg, 611209 |
| COG4 | 100 | 100 | 100 | 100 | Congenital disorder of glycosylation, type IIj, 613489<br>Saul-Wilson syndrome, 618150 |
| COG5 | 99,1 | 96,8 | 100 | 100 | Congenital disorder of glycosylation, type IIi, 613612 |

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| COG6 | 98,5 | 93,1 | 100 | 100 | Shaheen syndrome, 615328<br>Congenital disorder of glycosylation, type IIl, 614576 |
| COG7 | 100 | 99,4 | 100 | 100 | Congenital disorder of glycosylation, type IIe, 608779 |
| COG8 | 98,6 | 95,3 | 100 | 100 | Congenital disorder of glycosylation, type IIh, 611182 |
| COL4A1 | 99 | 97 | 100 | 100 | ?Retinal arteries, tortuosity of, 180000<br>Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773<br>Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564<br>Brain small vessel disease with or without ocular anomalies, 175780 |
| COL4A2 | 100 | 99,6 | 100 | 100 | Brain small vessel disease 2, 614483 |
| COLEC11 | 100 | 100 | 100 | 100 | 3MC syndrome 2, 265050 |
| COPB1 | 98,4 | 94,5 | 100 | 99,9 | Baralle-Macken syndrome, 619255 |
| COQ2 | 97,6 | 96,7 | 97,2 | 97,2 | Coenzyme Q10 deficiency, primary, 1, 607426 |
| COQ4 | 91 | 89,7 | 100 | 100 | Coenzyme Q10 deficiency, primary, 7, 616276 |
| COQ8A | 100 | 99,6 | 100 | 100 | Coenzyme Q10 deficiency, primary, 4, 612016 |
| COQ9 | 100 | 98,7 | 100 | 100 | Coenzyme Q10 deficiency, primary, 5, 614654 |
| COX10 | 100 | 99,9 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 3, 619046 |
| COX15 | 99,9 | 97,8 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 6, 615119 |
| COX16 | 99,3 | 96,9 | 100 | 99,9 | Mitochondrial complex IV deficiency, nuclear type 22, 619355 |
| COX6B1 | 100 | 100 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 7, 619051 |
| CPE | 99,8 | 98,8 | 100 | 100 | Intellectual developmental disorder and hypogonadotropic hypogonadism, 619326 |
| CPLANE1 | 99,4 | 98,2 | 100 | 100 | Orofaciodigital syndrome VI, 277170<br>Joubert syndrome 17, 614615 |
| CPLX1 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 63, 617976 |
| CPS1 | 100 | 100 | 100 | 100 | Carbamoylphosphate synthetase I deficiency, 237300 |
| CRADD | 99,9 | 97,5 | 100 | 100 | Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 |
| CRBN | 87,9 | 87,8 | 96,3 | 91,8 | Mental retardation, autosomal recessive 2, 607417 |
| CREBBP | 99,6 | 97,8 | 100 | 100 | Menke-Hennekam syndrome 1, 618332<br>Rubinstein-Taybi syndrome 1, 180849 |
| CRLF1 | 91,1 | 90,3 | 97,9 | 95,7 | Cold-induced sweating syndrome 1, 272430 |
| CRPPA | 98,4 | 94,7 | 100 | 99,8 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 |
| CSDE1 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |

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|---|---|---|---|---|---|
| CSF1R | 100 | 99,6 | 100 | 100 | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476<br>Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 |
| CSNK1G1 | 98,4 | 97,6 | 100 | 100 | No OMIM disease ID |
| CSNK2A1 | 81,1 | 76,6 | 94 | 94 | Okur-Chung neurodevelopmental syndrome, 617062 |
| CSNK2B | 100 | 100 | 100 | 100 | Poirier-Bienvenu neurodevelopmental syndrome, 618732 |
| CSPP1 | 99,7 | 98,1 | 100 | 100 | Joubert syndrome 21, 615636 |
| CSTB | 99,6 | 90,5 | 100 | 100 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 |
| CTBP1 | 94,3 | 86,9 | 99,4 | 98,4 | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 |
| CTC1 | 100 | 99,1 | 100 | 100 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| CTCF | 99,7 | 98,4 | 100 | 100 | Mental retardation, autosomal dominant 21, 615502 |
| CTDP1 | 88,7 | 85 | 100 | 99,8 | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 |
| CTNNA2 | 99,9 | 99,7 | 100 | 100 | Cortical dysplasia, complex, with other brain malformations 9, 618174 |
| CTNNB1 | 100 | 100 | 100 | 100 | Exudative vitreoretinopathy 7, 617572<br>Pilomatricoma, somatic, 132600<br>Colorectal cancer, somatic, 114500<br>Neurodevelopmental disorder with spastic diplegia and visual defects, 615075<br>Medulloblastoma, somatic, 155255<br>Ovarian cancer, somatic, 167000<br>Hepatocellular carcinoma, somatic, 114550 |
| CTNND1 | 100 | 99,8 | 100 | 100 | Blepharocheilodontic syndrome 2, 617681 |
| CTNND2 | 93,7 | 89,7 | 97,3 | 95,2 | No OMIM disease ID |
| CTSA | 100 | 99,6 | 100 | 100 | Galactosialidosis, 256540 |
| CTSD | 98,4 | 95 | 100 | 100 | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTTNBP2 | 99,3 | 96,4 | 100 | 100 | No OMIM disease ID |
| CTU2 | 100 | 98,7 | 100 | 100 | Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 |
| CUL3 | 99,4 | 97,4 | 100 | 100 | Neurodevelopmental disorder with or without autism or seizures, 619239<br>Pseudohypoaldosteronism, type IIE, 614496 |
| CUL4B | 96,9 | 88,8 | 99,9 | 98,5 | Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 |
| CUX1 | 96,5 | 94,6 | 99,5 | 98,7 | Global developmental delay with or without impaired intellectual development, 618330 |
| CUX2 | 99,9 | 99,3 | 100 | 100 | Developmental and epileptic encephalopathy 67, 618141 |
| CWC27 | 99,5 | 95,9 | 100 | 100 | Retinitis pigmentosa with or without skeletal anomalies, 250410 |

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|---|---|---|---|---|---|
| CWF19L1 | 100 | 99,6 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 17, 616127 |
| CYB5R3 | 99,1 | 98,1 | 99,6 | 98,5 | Methemoglobinemia, type I, 250800<br>Methemoglobinemia, type II, 250800 |
| CYFIP2 | 99,9 | 98,7 | 100 | 100 | Developmental and epileptic encephalopathy 65, 618008 |
| CYP27A1 | 99,7 | 98,1 | 100 | 100 | Cerebrotendinous xanthomatosis, 213700 |
| CYP2U1 | 95,3 | 92 | 100 | 99,9 | Spastic paraplegia 56, autosomal recessive, 615030 |
| D2HGDH | 99,7 | 98,2 | 100 | 100 | D-2-hydroxyglutaric aciduria, 600721 |
| DAG1 | 100 | 99,9 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538<br>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 |
| DARS1 | 99,5 | 99,3 | 100 | 99,9 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 |
| DARS2 | 94,8 | 93,8 | 100 | 100 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 |
| DBT | 99,1 | 96,1 | 100 | 100 | Maple syrup urine disease, type II, 248600 |
| DCAF17 | 98,5 | 93,4 | 100 | 100 | Woodhouse-Sakati syndrome, 241080 |
| DCC | 100 | 100 | 100 | 100 | Mirror movements 1 and/or agenesis of the corpus callosum, 157600<br>Esophageal carcinoma, somatic, 133239<br>Colorectal cancer, somatic, 114500<br>Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 |
| DCHS1 | 99,9 | 99,4 | 100 | 100 | Mitral valve prolapse 2, 607829<br>Van Maldergem syndrome 1, 601390 |
| DCPS | 91,3 | 91,2 | 100 | 100 | Al-Raqad syndrome, 616459 |
| DCX | 100 | 98,9 | 100 | 100 | Subcortical laminal heterotopia, X-linked, 300067<br>Lissencephaly, X-linked, 300067 |
| DDB1 | 100 | 99,5 | 100 | 100 | White-Kernohan syndrome, 619426 |
| DDC | 99,2 | 95 | 100 | 100 | Aromatic L-amino acid decarboxylase deficiency, 608643 |
| DDHD2 | 99,7 | 99,5 | 100 | 100 | Spastic paraplegia 54, autosomal recessive, 615033 |
| DDX11 | 84,9 | 80 | 100 | 100 | Warsaw breakage syndrome, 613398 |
| DDX23 | 99,7 | 97,6 | 100 | 100 | No OMIM disease ID |
| DDX3X | 81,1 | 78,6 | 98,3 | 96,1 | Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958 |
| DDX59 | 100 | 99,8 | 100 | 100 | Orofaciodigital syndrome V, 174300 |
| DDX6 | 95,8 | 81,5 | 100 | 100 | Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 |

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| DEAF1 | 99 | 94,5 | 99,9 | 98,2 | Vulto-van Silfout-de Vries syndrome, 615828<br>Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 |
| DEGS1 | 100 | 100 | 100 | 100 | Leukodystrophy, hypomyelinating, 18, 618404 |
| DENND5A | 99,8 | 98,7 | 100 | 100 | Developmental and epileptic encephalopathy 49, 617281 |
| DEPDC5 | 99,9 | 99,7 | 100 | 100 | Epilepsy, familial focal, with variable foci 1, 604364 |
| DHCR24 | 97,7 | 97,7 | 97,7 | 97,7 | Desmosterolosis, 602398 |
| DHCR7 | 100 | 100 | 100 | 100 | Smith-Lemli-Opitz syndrome, 270400 |
| DHDDS | 99,4 | 95,6 | 95,2 | 95,2 | Developmental delay and seizures with or without movement abnormalities, 617836<br>?Congenital disorder of glycosylation, type 1bb, 613861<br>Retinitis pigmentosa 59, 613861 |
| DHFR | 88,9 | 76,3 | 100 | 100 | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 |
| DHPS | 100 | 99,8 | 93,3 | 93,2 | Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 |
| DHTKD1 | 99,8 | 98,8 | 100 | 100 | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025<br>Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 |
| DHX16 | 100 | 99,7 | 100 | 100 | Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 |
| DHX30 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with severe motor impairment and absent language, 617804 |
| DHX37 | 99,8 | 97 | 100 | 100 | Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731<br>46, XY sex reversal 11, 273250 |
| DIAPH1 | 99,8 | 98,4 | 99,9 | 99 | Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900<br>Seizures, cortical blindness, microcephaly syndrome, 616632 |
| DIP2B | 99,8 | 99 | 100 | 100 | Mental retardation, FRA12A type, 136630 |
| DIS3L2 | 100 | 100 | 100 | 100 | Perlman syndrome, 267000 |
| DKC1 | 99,7 | 97,2 | 100 | 99,6 | Dyskeratosis congenita, X-linked, 305000 |
| DLD | 99,9 | 99,7 | 100 | 99,9 | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| DLG3 | 98,9 | 92,5 | 100 | 100 | Intellectual developmental disorder, X-linked 90, 300850 |
| DLG4 | 99,1 | 98,7 | 98,8 | 98,8 | Intellectual developmental disorder 62, 618793 |
| DLL1 | 100 | 98,8 | 100 | 100 | Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 |
| DMD | 99,5 | 98,1 | 100 | 99,9 | Becker muscular dystrophy, 300376<br>Cardiomyopathy, dilated, 3B, 302045<br>Duchenne muscular dystrophy, 310200 |
| DMPK | 99,8 | 98,4 | 100 | 100 | Myotonic dystrophy 1, 160900 |

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| DMXL2 | 99,7 | 98,9 | 100 | 99,9 | Developmental and epileptic encephalopathy 81, 618663<br>?Deafness, autosomal dominant 71, 617605<br>?Polyendocrine-polyneuropathy syndrome, 616113 |
| DNAJC12 | 87,4 | 87,3 | 100 | 100 | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 |
| DNAJC19 | 99,3 | 92,2 | 100 | 100 | 3-methylglutaconic aciduria, type V, 610198 |
| DNM1 | 92,7 | 89 | 97,5 | 97,4 | Developmental and epileptic encephalopathy 31, 616346 |
| DNM1L | 99,6 | 98,3 | 100 | 100 | Optic atrophy 5, 610708<br>Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 |
| DNMT3A | 99,8 | 98,2 | 100 | 100 | Tatton-Brown-Rahman syndrome, 615879<br>Acute myeloid leukemia, somatic, 601626<br>Heyn-Sproul-Jackson syndrome, 618724 |
| DNMT3B | 100 | 99,9 | 100 | 100 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860<br>Facioscapulohumeral muscular dystrophy 4, digenic, 619478 |
| DOCK3 | 99,9 | 98,9 | 100 | 100 | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 |
| DOCK6 | 99,4 | 98,7 | 100 | 100 | Adams-Oliver syndrome 2, 614219 |
| DOCK7 | 99,6 | 98,3 | 100 | 99,9 | Developmental and epileptic encephalopathy 23, 615859 |
| DOLK | 100 | 100 | 100 | 100 | Congenital disorder of glycosylation, type Im, 610768 |
| DONSON | 93,8 | 85,8 | 100 | 100 | Microcephaly, short stature, and limb abnormalities, 617604<br>Microcephaly-micromelia syndrome, 251230 |
| DPAGT1 | 100 | 99,8 | 100 | 100 | Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750<br>Congenital disorder of glycosylation, type Ij, 608093 |
| DPF2 | 99,5 | 96,4 | 100 | 100 | Coffin-Siris syndrome 7, 618027 |
| DPH1 | 100 | 99,9 | 100 | 100 | Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 |
| DPM1 | 97,4 | 90,9 | 98,6 | 94,6 | Congenital disorder of glycosylation, type Ie, 608799 |
| DPM2 | 100 | 97,7 | 100 | 100 | Congenital disorder of glycosylation, type Iu, 615042 |
| DPP6 | 99,7 | 97,9 | 98,8 | 96,8 | Mental retardation, autosomal dominant 33, 616311 |
| DPYD | 99,5 | 96,5 | 100 | 100 | Dihydropyrimidine dehydrogenase deficiency, 274270<br>5-fluorouracil toxicity, 274270 |
| DPYS | 100 | 100 | 100 | 100 | Dihydropyrimidinuria, 222748 |
| DPYSL5 | 100 | 99,9 | 100 | 100 | Ritscher-Schinzel syndrome 4, 619435 |
| DYM | 97 | 95,6 | 100 | 100 | Smith-McCort dysplasia, 607326<br>Dyggve-Melchior-Clausen disease, 223800 |

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|---|---|---|---|---|---|
| DYNC1H1 | 99,9 | 99,3 | 100 | 100 | Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600<br>Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563 |
| DYNC1I2 | 84 | 66 | 100 | 100 | Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 |
| DYRK1A | 100 | 100 | 100 | 100 | Mental retardation, autosomal dominant 7, 614104 |
| EARS2 | 99,8 | 98 | 100 | 100 | Combined oxidative phosphorylation deficiency 12, 614924 |
| EBF3 | 100 | 100 | 100 | 100 | Hypotonia, ataxia, and delayed development syndrome, 617330 |
| EBP | 99,5 | 94,3 | 100 | 100 | MEND syndrome, 300960<br>Chondrodysplasia punctata, X-linked dominant, 302960 |
| ECHS1 | 100 | 99,4 | 100 | 100 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 |
| EDC3 | 100 | 99,5 | 100 | 100 | ?Mental retardation, autosomal recessive 50, 616460 |
| EED | 95,6 | 91,7 | 100 | 100 | Cohen-Gibson syndrome, 617561 |
| EEF1A2 | 100 | 100 | 100 | 99,4 | Mental retardation, autosomal dominant 38, 616393<br>Developmental and epileptic encephalopathy 33, 616409 |
| EFNB2 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| EFTUD2 | 100 | 99,2 | 100 | 100 | Mandibulofacial dysostosis, Guion-Almeida type, 610536 |
| EHMT1 | 94,5 | 93,6 | 99,6 | 99,5 | Kleefstra syndrome 1, 610253 |
| EIF2AK1 | 98,1 | 94,5 | 100 | 100 | ?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 |
| EIF2AK2 | 99,7 | 98,9 | 100 | 99,9 | Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 |
| EIF2AK3 | 98,2 | 95,5 | 100 | 99,8 | Wolcott-Rallison syndrome, 226980 |
| EIF2B4 | 100 | 99,5 | 100 | 100 | Ovarioleukodystrophy, 603896<br>Leukoencephalopathy with vanishing white matter, 603896 |
| EIF2B5 | 99,8 | 98,5 | 100 | 100 | Ovarioleukodystrophy, 603896<br>Leukoencephalopathy with vanishing white matter, 603896 |
| EIF2S3 | 95 | 86,8 | 100 | 100 | MEHMO syndrome, 300148 |
| EIF3F | 97,1 | 82,5 | 100 | 100 | Mental retardation, autosomal recessive 67, 618295 |
| EIF4A3 | 100 | 99,2 | 100 | 100 | Robin sequence with cleft mandible and limb anomalies, 268305 |
| EIF5A | 99,8 | 96,4 | 100 | 100 | Faundes-Banka syndrome, 619376 |
| ELAC2 | 100 | 99,2 | 100 | 100 | Combined oxidative phosphorylation deficiency 17, 615440 |
| ELOVL4 | 99,7 | 98,9 | 100 | 99,9 | Spinocerebellar ataxia 34, 133190<br>Stargardt disease 3, 600110<br>Ichthyosis, spastic quadriplegia, and mental retardation, 614457 |

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|---|---|---|---|---|---|
| ELP2 | 99,8 | 98,3 | 100 | 99,9 | Mental retardation, autosomal recessive 58, 617270 |
| EMC1 | 99,9 | 98 | 100 | 100 | Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 |
| EMC10 | 97,1 | 92 | 100 | 100 | Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 |
| EML1 | 99,6 | 98,1 | 100 | 100 | Band heterotopia, 600348 |
| EMX2 | 100 | 100 | 100 | 100 | Schizencephaly, 269160 |
| ENTPD1 | 100 | 99,8 | 100 | 100 | Spastic paraplegia 64, autosomal recessive, 615683 |
| EP300 | 99,9 | 98,9 | 100 | 100 | Menke-Hennekam syndrome 2, 618333<br>Colorectal cancer, somatic, 114500<br>Rubinstein-Taybi syndrome 2, 613684 |
| EPG5 | 99,2 | 97,8 | 100 | 100 | Vici syndrome, 242840 |
| EPHA7 | 100 | 99,4 | 100 | 100 | No OMIM disease ID |
| ERCC1 | 100 | 96,4 | 100 | 100 | Cerebrooculofacioskeletal syndrome 4, 610758 |
| ERCC2 | 100 | 99,4 | 100 | 100 | Xeroderma pigmentosum, group D, 278730<br>Trichothiodystrophy 1, photosensitive, 601675<br>?Cerebrooculofacioskeletal syndrome 2, 610756 |
| ERCC3 | 96,8 | 95,6 | 100 | 100 | Trichothiodystrophy 2, photosensitive, 616390<br>Xeroderma pigmentosum, group B, 610651 |
| ERCC5 | 99,9 | 99 | 100 | 100 | Xeroderma pigmentosum, group G, 278780<br>Cerebrooculofacioskeletal syndrome 3, 616570<br>Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 |
| ERCC6 | 100 | 100 | 100 | 100 | UV-sensitive syndrome 1, 600630<br>Cerebrooculofacioskeletal syndrome 1, 214150<br>Cockayne syndrome, type B, 133540<br>De Sanctis-Cacchione syndrome, 278800<br>Premature ovarian failure 11, 616946 |
| ERCC8 | 99 | 94,8 | 100 | 100 | UV-sensitive syndrome 2, 614621<br>Cockayne syndrome, type A, 216400 |
| ERLIN2 | 100 | 99,1 | 100 | 99,9 | Spastic paraplegia 18, autosomal recessive, 611225 |
| ESCO2 | 98,5 | 94,6 | 100 | 99,7 | Juberg-Hayward syndrome, 216100<br>Roberts-SC phocomelia syndrome, 268300 |
| ETFB | 100 | 99,9 | 100 | 100 | Glutaric acidemia IIB, 231680 |
| ETHE1 | 99,3 | 93,3 | 100 | 100 | Ethylmalonic encephalopathy, 602473 |
| EXOC2 | 99,8 | 99,5 | 100 | 100 | Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 |
| EXOC7 | 100 | 99,6 | 100 | 100 | Neurodevelopmental disorder with seizures and brain atrophy, 619072 |

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|---|---|---|---|---|---|
| EXOSC2 | 100 | 99,9 | 100 | 100 | Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 |
| EXOSC3 | 98,1 | 90,5 | 100 | 100 | Pontocerebellar hypoplasia, type 1B, 614678 |
| EXOSC8 | 98,7 | 90 | 100 | 100 | Pontocerebellar hypoplasia, type 1C, 616081 |
| EXOSC9 | 99,3 | 94,7 | 100 | 99,9 | Pontocerebellar hypoplasia, type 1D, 618065 |
| EXTL3 | 100 | 100 | 100 | 100 | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 |
| EZH2 | 99,7 | 98 | 100 | 100 | Weaver syndrome, 277590 |
| FA2H | 92,4 | 82,6 | 100 | 100 | Spastic paraplegia 35, autosomal recessive, 612319 |
| FAM126A | 99,5 | 99,4 | 100 | 100 | Leukodystrophy, hypomyelinating, 5, 610532 |
| FAM149B1 | 98,2 | 94,3 | 100 | 100 | Joubert syndrome 36, 618763 |
| FAM20C | 100 | 100 | 100 | 99,7 | Raine syndrome, 259775 |
| FAM50A | 99,8 | 97 | 100 | 99,6 | Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261 |
| FAR1 | 97,4 | 94 | 100 | 100 | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154<br>Cataracts, spastic paraparesis, and speech delay, 619338 |
| FARS2 | 100 | 100 | 100 | 100 | Combined oxidative phosphorylation deficiency 14, 614946<br>Spastic paraplegia 77, autosomal recessive, 617046 |
| FARSB | 98 | 92,9 | 100 | 100 | Rajab interstitial lung disease with brain calcifications 1, 613658 |
| FAT4 | 100 | 100 | 100 | 100 | Van Maldergem syndrome 2, 615546<br>Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 |
| FBRSL1 | 56,4 | 50,8 | 96,5 | 92,4 | No OMIM disease ID |
| FBXL3 | 100 | 100 | 100 | 100 | Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 |
| FBXL4 | 100 | 100 | 100 | 100 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 |
| FBXO11 | 97,3 | 91,2 | 100 | 100 | Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 |
| FBXO31 | 97 | 94,2 | 100 | 100 | ?Mental retardation, autosomal recessive 45, 615979 |
| FDFT1 | 98,5 | 96,7 | 100 | 100 | Squalene synthase deficiency, 618156 |
| FGD1 | 97,2 | 91,2 | 100 | 100 | Mental retardation, X-linked syndromic 16, 305400<br>Aarskog-Scott syndrome, 305400 |
| FGF12 | 100 | 99,1 | 100 | 100 | Developmental and epileptic encephalopathy 47, 617166 |
| FGF13 | 99,1 | 95,1 | 100 | 100 | Developmental and epileptic encephalopathy 90, 301058 |
| FGF14 | 100 | 100 | 100 | 100 | Spinocerebellar ataxia 27, 609307 |
| FGFR1 | 100 | 99,3 | 100 | 100 | Pfeiffer syndrome, 101600<br>Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 |

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|---|---|---|---|---|---|
| | | | | | Jackson-Weiss syndrome, 123150<br>Hartsfield syndrome, 615465<br>Trigonocephaly 1, 190440<br>Osteoglophonic dysplasia, 166250<br>Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 |
| FGFR2 | 97,6 | 97 | 100 | 100 | Bent bone dysplasia syndrome, 614592<br>LADD syndrome, 149730<br>Scaphocephaly, maxillary retrusion, and mental retardation, 609579<br>Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410<br>Jackson-Weiss syndrome, 123150<br>Gastric cancer, somatic, 613659<br>Craniofacial-skeletal-dermatologic dysplasia, 101600<br>Apert syndrome, 101200<br>Pfeiffer syndrome, 101600<br>Beare-Stevenson cutis gyrata syndrome, 123790<br>Crouzon syndrome, 123500<br>Saethre-Chotzen syndrome, 101400<br>Scaphocephaly and Axenfeld-Rieger anomaly,<br>Craniosynostosis, nonspecific, |
| FGFR3 | 99,8 | 98 | 100 | 100 | Muenke syndrome, 602849<br>SADDAN, 616482<br>Hypochondroplasia, 146000<br>LADD syndrome, 149730<br>Thanatophoric dysplasia, type II, 187601<br>Nevus, epidermal, somatic, 162900<br>CATSHL syndrome, 610474<br>Thanatophoric dysplasia, type I, 187600<br>Spermatocytic seminoma, somatic, 273300<br>Bladder cancer, somatic, 109800<br>Achondroplasia, 100800<br>Cervical cancer, somatic, 603956<br>Colorectal cancer, somatic, 114500<br>Crouzon syndrome with acanthosis nigricans, 612247 |
| FH | 93,2 | 87,2 | 100 | 100 | Leiomyomatosis and renal cell cancer, 150800<br>Fumarase deficiency, 606812 |
| FIBP | 100 | 99,8 | 100 | 100 | Thauvin-Robinet-Faivre syndrome, 617107 |
| FIGN | 100 | 100 | 100 | 100 | No OMIM disease ID |

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|---|---|---|---|---|---|
| FKRP | 100 | 100 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612<br>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 |
| FKTN | 99,8 | 95,2 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800<br>Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152<br>Cardiomyopathy, dilated, 1X, 611615 |
| FLNA | 100 | 99,9 | 100 | 100 | Otopalatodigital syndrome, type II, 304120<br>Intestinal pseudoobstruction, neuronal, 300048<br>Cardiac valvular dysplasia, X-linked, 314400<br>?FG syndrome 2, 300321<br>Melnick-Needles syndrome, 309350<br>Terminal osseous dysplasia, 300244<br>Congenital short bowel syndrome, 300048<br>Otopalatodigital syndrome, type I, 311300<br>Heterotopia, periventricular, 1, 300049<br>Frontometaphyseal dysplasia 1, 305620 |
| FLVCR1 | 99,7 | 98,3 | 100 | 99,9 | Ataxia, posterior column, with retinitis pigmentosa, 609033 |
| FLVCR2 | 100 | 100 | 100 | 100 | Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 |
| FMN2 | 85,5 | 82,8 | 100 | 100 | Mental retardation, autosomal recessive 47, 616193 |
| FMR1 | 95,2 | 90,1 | 100 | 99,9 | Fragile X tremor/ataxia syndrome, 300623<br>Fragile X syndrome, 300624<br>Premature ovarian failure 1, 311360 |
| FOLR1 | 100 | 99,9 | 100 | 100 | Neurodegeneration due to cerebral folate transport deficiency, 613068 |
| FOXG1 | 87,2 | 79,4 | 98,3 | 94,8 | Rett syndrome, congenital variant, 613454 |
| FOXJ1 | 100 | 98,9 | 100 | 100 | Ciliary dyskinesia, primary, 43, 618699 |
| FOXP1 | 99,9 | 99,1 | 100 | 100 | Mental retardation with language impairment and with or without autistic features, 613670 |
| FOXP2 | 99,3 | 98,7 | 100 | 100 | Speech-language disorder-1, 602081 |
| FOXRED1 | 100 | 99,6 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 19, 618241 |
| FRAS1 | 100 | 99,2 | 100 | 100 | Fraser syndrome 1, 219000 |
| FRMD4A | 91,3 | 89,4 | 96,6 | 96,6 | ?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 |
| FRMPD4 | 97 | 94,2 | 98,3 | 98,2 | Intellectual developmental disorder, X-linked 104, 300983 |
| FRRS1L | 82,5 | 73,7 | 98,8 | 93,9 | Developmental and epileptic encephalopathy 37, 616981 |

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|---|---|---|---|---|---|
| FTCD | 97,7 | 93,2 | 100 | 100 | Glutamate formiminotransferase deficiency, 229100 |
| FTO | 83,8 | 83,7 | 94,2 | 94,2 | Growth retardation, developmental delay, facial dysmorphism, 612938 |
| FTSJ1 | 98,3 | 93,8 | 100 | 100 | Intellectual developmental disorder, X-linked 9, 309549 |
| FUCA1 | 100 | 100 | 100 | 100 | Fucosidosis, 230000 |
| FUT8 | 99,8 | 98,9 | 100 | 100 | Congenital disorder of glycosylation with defective fucosylation 1, 618005 |
| GABBR2 | 96,1 | 90,9 | 98,8 | 98 | Developmental and epileptic encephalopathy 59, 617904<br>Neurodevelopmental disorder with poor language and loss of hand skills, 617903 |
| GABRA1 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 19, 615744 |
| GABRA2 | 99,5 | 96,9 | 100 | 100 | Developmental and epileptic encephalopathy 78, 618557 |
| GABRA3 | 98 | 94 | 100 | 99,3 | No OMIM disease ID |
| GABRA5 | 100 | 99,4 | 100 | 100 | Developmental and epileptic encephalopathy 79, 618559 |
| GABRB1 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 45, 617153 |
| GABRB2 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 92, 617829 |
| GABRB3 | 99,8 | 98,1 | 100 | 100 | Developmental and epileptic encephalopathy 43, 617113 |
| GABRG2 | 89,9 | 88,4 | 93 | 93 | Developmental and epileptic encephalopathy 74, 618396<br>Febrile seizures, familial, 8, 607681<br>Generalized epilepsy with febrile seizures plus, type 3, 607681 |
| GAD1 | 100 | 99,3 | 100 | 100 | Developmental and epileptic encephalopathy 89, 619124 |
| GALC | 99,7 | 97,6 | 100 | 100 | Krabbe disease, 245200 |
| GALE | 100 | 100 | 100 | 100 | Galactose epimerase deficiency, 230350 |
| GALNT2 | 99,8 | 97,1 | 100 | 100 | Congenital disorder of glycosylation, type IIt, 618885 |
| GALT | 100 | 99,6 | 100 | 100 | Galactosemia, 230400 |
| GAMT | 95 | 82,7 | 100 | 100 | Cerebral creatine deficiency syndrome 2, 612736 |
| GATAD2B | 100 | 99,1 | 100 | 100 | GAND syndrome, 615074 |
| GATM | 100 | 100 | 100 | 100 | Cerebral creatine deficiency syndrome 3, 612718<br>Fanconi renotubular syndrome 1, 134600 |
| GCH1 | 99,9 | 97,3 | 100 | 100 | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230<br>Hyperphenylalaninemia, BH4-deficient, B, 233910 |
| GCSH | 75,7 | 64,4 | 100 | 100 | ?Glycine encephalopathy, 605899 |
| GDI1 | 99,8 | 98,6 | 100 | 100 | Intellectual developmental disorder, X-linked 41, 300849 |
| GEMIN5 | 99,9 | 98,7 | 100 | 100 | Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 |

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| GFAP | 91,7 | 89,5 | 100 | 100 | Alexander disease, 203450 |
| GFER | 99,8 | 97,6 | 100 | 100 | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 |
| GFM1 | 99,7 | 98,7 | 100 | 100 | Combined oxidative phosphorylation deficiency 1, 609060 |
| GFM2 | 98,1 | 93,7 | 100 | 100 | Combined oxidative phosphorylation deficiency 39, 618397 |
| GIGYF1 | 99,1 | 95,1 | 100 | 100 | No OMIM disease ID |
| GJA1 | 100 | 100 | 100 | 100 | Erythrokeratodermia variabilis et progressiva 3, 617525<br>Craniometaphyseal dysplasia, autosomal recessive, 218400<br>Oculodentodigital dysplasia, 164200<br>Hypoplastic left heart syndrome 1, 241550<br>Palmoplantar keratoderma with congenital alopecia, 104100<br>Syndactyly, type III, 186100<br>Oculodentodigital dysplasia, autosomal recessive, 257850<br>Atrioventricular septal defect 3, 600309 |
| GJB1 | 100 | 100 | 100 | 100 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 |
| GJC2 | 82,3 | 64,5 | 97,8 | 93,2 | Lymphatic malformation 3, 613480<br>Spastic paraplegia 44, autosomal recessive, 613206<br>Leukodystrophy, hypomyelinating, 2, 608804 |
| GK | 84,2 | 61,8 | 100 | 99,6 | Glycerol kinase deficiency, 307030 |
| GLB1 | 99,2 | 92,8 | 100 | 100 | GM1-gangliosidosis, type I, 230500<br>GM1-gangliosidosis, type III, 230650<br>Mucopolysaccharidosis type IVB (Morquio), 253010<br>GM1-gangliosidosis, type II, 230600 |
| GLDC | 88,9 | 77,8 | 100 | 99,9 | Glycine encephalopathy, 605899 |
| GLI2 | 99,8 | 98,6 | 100 | 99,9 | Culler-Jones syndrome, 615849<br>Holoprosencephaly 9, 610829 |
| GLI3 | 98,5 | 97,7 | 100 | 100 | Greig cephalopolysyndactyly syndrome, 175700<br>Polydactyly, postaxial, types A1 and B, 174200<br>Pallister-Hall syndrome, 146510<br>Polydactyly, preaxial, type IV, 174700 |
| GLIS3 | 98,5 | 97,4 | 100 | 100 | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 |
| GLS | 96,9 | 88,5 | 100 | 99,9 | Global developmental delay, progressive ataxia, and elevated glutamine, 618412<br>?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339<br>Developmental and epileptic encephalopathy 71, 618328 |
| GLUD1 | 96,4 | 84,4 | 100 | 100 | Hyperinsulinism-hyperammonemia syndrome, 606762 |
| GLYCTK | 98,7 | 97,3 | 100 | 100 | D-glyceric aciduria, 220120 |

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| GM2A | 100 | 100 | 100 | 100 | GM2-gangliosidosis, AB variant, 272750 |
| GMPPA | 100 | 100 | 100 | 100 | Alacrima, achalasia, and mental retardation syndrome, 615510 |
| GMPPB | 100 | 100 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 |
| GNAI1 | 97,2 | 89,7 | 100 | 100 | No OMIM disease ID |
| GNAO1 | 93,8 | 93,8 | 100 | 100 | Developmental and epileptic encephalopathy 17, 615473<br>Neurodevelopmental disorder with involuntary movements, 617493 |
| GNAS | 86,7 | 84,4 | 81,8 | 81,7 | ACTH-independent macronodular adrenal hyperplasia, 219080<br>Pituitary adenoma 3, multiple types, somatic, 617686<br>Pseudohypoparathyroidism Ic, 612462<br>Pseudohypoparathyroidism Ia, 103580<br>Osseous heteroplasia, progressive, 166350<br>Pseudohypoparathyroidism Ib, 603233<br>McCune-Albright syndrome, somatic, mosaic, 174800<br>Pseudopseudohypoparathyroidism, 612463 |
| GNB1 | 100 | 100 | 100 | 100 | Myelodysplastic syndrome, somatic, 614286<br>Leukemia, acute lymphoblastic, somatic, 613065<br>Mental retardation, autosomal dominant 42, 616973 |
| GNB5 | 99,9 | 96,5 | 100 | 100 | Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182<br>Intellectual developmental disorder with cardiac arrhythmia, 617173 |
| GNPAT | 99,5 | 95,6 | 100 | 100 | Rhizomelic chondrodysplasia punctata, type 2, 222765 |
| GNPTAB | 99,9 | 99,7 | 100 | 100 | Mucolipidosis III alpha/beta, 252600<br>Mucolipidosis II alpha/beta, 252500 |
| GNPTG | 99,8 | 96,6 | 100 | 100 | Mucolipidosis III gamma, 252605 |
| GNS | 99,2 | 94,6 | 100 | 100 | Mucopolysaccharidosis type IIID, 252940 |
| GOT2 | 94,6 | 87 | 100 | 100 | Developmental and epileptic encephalopathy 82, 618721 |
| GPAA1 | 98,6 | 95,5 | 100 | 100 | Glycosylphosphatidylinositol biosynthesis defect 15, 617810 |
| GPC3 | 98,8 | 92,9 | 100 | 99,9 | Wilms tumor, somatic, 194070<br>Simpson-Golabi-Behmel syndrome, type 1, 312870 |
| GPC4 | 99,9 | 98,3 | 100 | 100 | Keipert syndrome, 301026 |
| GPHN | 99,9 | 99,1 | 100 | 100 | Molybdenum cofactor deficiency C, 615501 |
| GPSM2 | 99,9 | 99,3 | 100 | 100 | Chudley-McCullough syndrome, 604213 |
| GPT2 | 99,4 | 95,3 | 100 | 100 | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 |

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| GRIA2 | 99,7 | 96,6 | 100 | 100 | Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 |
| GRIA3 | 99,5 | 94 | 99,9 | 98,6 | Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699 |
| GRIA4 | 99,8 | 98,9 | 100 | 100 | Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 |
| GRID2 | 100 | 99,8 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 18, 616204 |
| GRIK2 | 96,1 | 95,3 | 96,3 | 96,3 | Mental retardation, autosomal recessive, 6, 611092 |
| GRIN1 | 100 | 99,9 | 100 | 100 | Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820<br>Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 |
| GRIN2A | 100 | 100 | 100 | 100 | Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 |
| GRIN2B | 99,6 | 98,7 | 100 | 100 | Developmental and epileptic encephalopathy 27, 616139<br>Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 |
| GRIN2D | 83,8 | 69,4 | 93,7 | 87,4 | Developmental and epileptic encephalopathy 46, 617162 |
| GRIP1 | 100 | 99,3 | 100 | 100 | Fraser syndrome 3, 617667 |
| GRM1 | 100 | 99,5 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 13, 614831<br>Spinocerebellar ataxia 44, 617691 |
| GRM7 | 99,9 | 99,1 | 100 | 100 | Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 |
| GRN | 100 | 100 | 100 | 100 | Aphasia, primary progressive, 607485<br>Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485<br>Ceroid lipofuscinosis, neuronal, 11, 614706 |
| GSE1 | 100 | 99,4 | 100 | 100 | No OMIM disease ID |
| GSS | 96,5 | 96,3 | 100 | 100 | Hemolytic anemia due to glutathione synthetase deficiency, 231900<br>Glutathione synthetase deficiency, 266130 |
| GTF2H5 | 72,2 | 71,7 | 72,5 | 72,5 | Trichothiodystrophy 3, photosensitive, 616395 |
| GTPBP2 | 99,8 | 98,5 | 100 | 100 | Jaberi-Elahi syndrome, 617988 |
| GTPBP3 | 100 | 99,9 | 100 | 100 | Combined oxidative phosphorylation deficiency 23, 616198 |
| GUSB | 92,5 | 90,1 | 100 | 100 | Mucopolysaccharidosis VII, 253220 |
| H1-4 | 100 | 100 | 100 | 100 | Rahman syndrome, 617537 |
| H4C3 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| HACE1 | 99,7 | 99,3 | 100 | 99,9 | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 |
| HADH | 99,2 | 97,7 | 100 | 100 | Hyperinsulinemic hypoglycemia, familial, 4, 609975<br>3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 |

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| HADHA | 95,5 | 88,3 | 100 | 100 | HELLP syndrome, maternal, of pregnancy, 609016<br>Mitochondrial trifunctional protein deficiency, 609015<br>LCHAD deficiency, 609016<br>Fatty liver, acute, of pregnancy, 609016 |
| HADHB | 97,7 | 87 | 100 | 99,9 | Trifunctional protein deficiency, 609015 |
| HAX1 | 100 | 100 | 100 | 100 | Neutropenia, severe congenital 3, autosomal recessive, 610738 |
| HCCS | 99,3 | 96,1 | 100 | 100 | Linear skin defects with multiple congenital anomalies 1, 309801 |
| HCFC1 | 98,1 | 93 | 100 | 100 | Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type ), 309541 |
| HCN1 | 98,4 | 98,3 | 98,5 | 98,4 | Developmental and epileptic encephalopathy 24, 615871<br>Generalized epilepsy with febrile seizures plus, type 10, 618482 |
| HDAC4 | 100 | 99,9 | 100 | 100 | No OMIM disease ID |
| HDAC6 | 99,5 | 97,1 | 100 | 99,9 | ?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 |
| HDAC8 | 85,7 | 83,7 | 96,4 | 95,2 | Cornelia de Lange syndrome 5, 300882 |
| HEATR5B | 100 | 99,3 | 100 | 100 | No OMIM disease ID |
| HECW2 | 99,8 | 98,2 | 100 | 100 | Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 |
| HEPACAM | 86,8 | 78,5 | 100 | 100 | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925<br>Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 |
| HERC1 | 100 | 99,9 | 100 | 100 | Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 |
| HERC2 | 79,7 | 76,7 | 100 | 100 | Mental retardation, autosomal recessive 38, 615516 |
| HESX1 | 99,3 | 97,3 | 100 | 100 | Pituitary hormone deficiency, combined, 5, 182230<br>Septooptic dysplasia, 182230<br>Growth hormone deficiency with pituitary anomalies, 182230 |
| HEXA | 93,8 | 93,1 | 100 | 100 | GM2-gangliosidosis, several forms, 272800<br>Tay-Sachs disease, 272800 |
| HEXB | 99,4 | 96,6 | 100 | 100 | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |
| HGSNAT | 86,4 | 86,2 | 91,3 | 89,1 | Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930<br>Retinitis pigmentosa 73, 616544 |
| HIBCH | 98,2 | 84,5 | 100 | 100 | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 |
| HIVEP2 | 100 | 99,9 | 100 | 100 | Mental retardation, autosomal dominant 43, 616977 |
| HK1 | 100 | 99,9 | 100 | 100 | Retinitis pigmentosa 79, 617460<br>Neuropathy, hereditary motor and sensory, Russe type, 605285 |

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|---|---|---|---|---|---|
| | | | | | Neurodevelopmental disorder with visual defects and brain anomalies, 618547<br>Hemolytic anemia due to hexokinase deficiency, 235700 |
| HLCS | 100 | 100 | 100 | 100 | Holocarboxylase synthetase deficiency, 253270 |
| HMGCL | 100 | 99,4 | 100 | 100 | HMG-CoA lyase deficiency, 246450 |
| HNMT | 99,9 | 99,5 | 100 | 100 | Mental retardation, autosomal recessive 51, 616739 |
| HNRNPD | 86,3 | 79,8 | 100 | 100 | No OMIM disease ID |
| HNRNPH1 | 99,1 | 94,2 | 100 | 100 | No OMIM disease ID |
| HNRNPH2 | 100 | 100 | 100 | 100 | Intellectual developmental disorder, X-linked, syndromic, Bain type, 300986 |
| HNRNPK | 88,8 | 78,1 | 100 | 100 | Au-Kline syndrome, 616580 |
| HNRNPU | 99,9 | 98,7 | 100 | 100 | Developmental and epileptic encephalopathy 54, 617391 |
| HOXA1 | 100 | 100 | 100 | 100 | Bosley-Salih-Alorainy syndrome, 601536<br>Athabaskan brainstem dysgenesis syndrome, 601536 |
| HPD | 100 | 99,8 | 100 | 100 | Hawkinsinuria, 140350<br>Tyrosinemia, type III, 276710 |
| HPDL | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026<br>Spastic paraplegia 83, autosomal recessive, 619027 |
| HPRT1 | 98,6 | 90,6 | 99,5 | 98,4 | Hyperuricemia, HRPT-related, 300323<br>Lesch-Nyhan syndrome, 300322 |
| HRAS | 100 | 100 | 100 | 100 | Bladder cancer, somatic, 109800<br>Thyroid carcinoma, follicular, somatic, 188470<br>Congenital myopathy with excess of muscle spindles, 218040<br>Nevus sebaceous or woolly hair nevus, somatic, 162900<br>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200<br>Spitz nevus or nevus spilus, somatic, 137550<br>Costello syndrome, 218040 |
| HS2ST1 | 99,6 | 99,2 | 100 | 99,5 | Neurofacioskeletal syndrome with or without renal agenesis, 619194 |
| HSD17B10 | 99,9 | 98,3 | 100 | 100 | HSD10 mitochondrial disease, 300438 |
| HSD17B4 | 95,3 | 92,8 | 96,6 | 96,6 | D-bifunctional protein deficiency, 261515<br>Perrault syndrome 1, 233400 |
| HSPA9 | 87,1 | 82,8 | 100 | 100 | Even-plus syndrome, 616854<br>Anemia, sideroblastic, 4, 182170 |
| HSPD1 | 96,7 | 90 | 100 | 100 | Spastic paraplegia 13, autosomal dominant, 605280<br>Leukodystrophy, hypomyelinating, 4, 612233 |
| HTRA2 | 100 | 99,6 | 100 | 100 | 3-methylglutaconic aciduria, type VIII, 617248 |

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|---|---|---|---|---|---|
| HUWE1 | 98,6 | 93,2 | 100 | 100 | Intellectual developmental disorder, X-linked, Turner type, 309590 |
| HYLS1 | 100 | 100 | 100 | 100 | Hydrolethalus syndrome, 236680 |
| IARS1 | 99,9 | 99,4 | 100 | 100 | Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 |
| IARS2 | 99,9 | 99,8 | 100 | 100 | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 |
| IBA57 | 95,4 | 91,7 | 100 | 100 | Multiple mitochondrial dysfunctions syndrome 3, 615330<br>?Spastic paraplegia 74, autosomal recessive, 616451 |
| IDS | 99,6 | 95,3 | 100 | 100 | Mucopolysaccharidosis II, 309900 |
| IDUA | 94,6 | 87,4 | 100 | 100 | Mucopolysaccharidosis Is, 607016<br>Mucopolysaccharidosis Ih/s, 607015<br>Mucopolysaccharidosis Ih, 607014 |
| IER3IP1 | 92 | 80,2 | 100 | 100 | Microcephaly, epilepsy, and diabetes syndrome, 614231 |
| IFIH1 | 99,5 | 97,3 | 100 | 100 | Aicardi-Goutieres syndrome 7, 615846<br>Singleton-Merten syndrome 1, 182250 |
| IFT172 | 99,6 | 98,6 | 100 | 100 | Retinitis pigmentosa 71, 616394<br>Bardet-Biedl syndrome 20, 619471<br>Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 |
| IFT27 | 100 | 100 | 100 | 100 | Bardet-Biedl syndrome 19, 615996 |
| IFT81 | 92,9 | 89,6 | 94,9 | 94,6 | Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 |
| IGBP1 | 98,8 | 93,5 | 100 | 100 | ?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 |
| IGF1 | 99,8 | 99,8 | 100 | 100 | Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 |
| IGF1R | 100 | 99,7 | 100 | 100 | Insulin-like growth factor I, resistance to, 270450 |
| IKBKG | 84,6 | 75,2 | 100 | 100 | Incontinentia pigmenti, 308300<br>Ectodermal dysplasia and immunodeficiency 1, 300291<br>Immunodeficiency 33, 300636 |
| IL1RAPL1 | 99,8 | 98,5 | 100 | 100 | Intellectual developmental disorder, X-linked 21, 300143 |
| IMPA1 | 96,1 | 86,4 | 100 | 99,8 | Mental retardation, autosomal recessive 59, 617323 |
| INPP5E | 96,9 | 93,2 | 100 | 100 | Joubert syndrome 1, 213300<br>Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 |
| INPP5K | 100 | 99,7 | 100 | 100 | Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 |
| INTS1 | 99,8 | 98,6 | 100 | 100 | Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 |
| IQSEC1 | 88,6 | 86,1 | 97,6 | 94,8 | Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 |

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|---|---|---|---|---|---|
| IQSEC2 | 94,6 | 84 | 99,5 | 98,3 | Intellectual developmental disorder, X-linked 1, 309530 |
| IREB2 | 99,9 | 99,8 | 100 | 100 | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 |
| IRF2BPL | 100 | 97,9 | 99,9 | 99 | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 |
| ISCA2 | 99,8 | 96,5 | 100 | 100 | Multiple mitochondrial dysfunctions syndrome 4, 616370 |
| ITGA7 | 99,7 | 97,9 | 100 | 100 | Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 |
| ITPA | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 35, 616647 |
| ITPR1 | 100 | 99,5 | 100 | 100 | Gillespie syndrome, 206700<br>Spinocerebellar ataxia 29, congenital nonprogressive, 117360<br>Spinocerebellar ataxia 15, 606658 |
| IVD | 100 | 99,9 | 100 | 100 | Isovaleric acidemia, 243500 |
| JAG1 | 97,8 | 96,7 | 100 | 100 | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992<br>Alagille syndrome 1, 118450<br>Tetralogy of Fallot, 187500 |
| JAM3 | 100 | 100 | 100 | 100 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 |
| JARID2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| JMJD1C | 99,5 | 98,9 | 100 | 100 | No OMIM disease ID |
| KANK1 | 100 | 99,9 | 100 | 100 | Cerebral palsy, spastic quadriplegic, 2, 612900 |
| KANSL1 | 99,8 | 98,2 | 100 | 100 | Koolen-De Vries syndrome, 610443 |
| KAT5 | 99,8 | 97,8 | 100 | 100 | Neurodevelopmental disorder wtih dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 |
| KAT6A | 100 | 99,2 | 100 | 100 | Arboleda-Tham syndrome, 616268 |
| KAT6B | 99,4 | 98 | 100 | 100 | SBBYSS syndrome, 603736<br>Genitopatellar syndrome, 606170 |
| KAT8 | 100 | 98,6 | 100 | 100 | Li-Ghorgani-Weisz-Hubshman syndrome, 618974 |
| KATNB1 | 100 | 100 | 100 | 100 | Lissencephaly 6, with microcephaly, 616212 |
| KCNA2 | 100 | 99,6 | 100 | 100 | Developmental and epileptic encephalopathy 32, 616366 |
| KCNA4 | 100 | 100 | 100 | 100 | Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 |
| KCNB1 | 100 | 99,7 | 100 | 100 | Developmental and epileptic encephalopathy 26, 616056 |
| KCNC1 | 100 | 100 | 100 | 100 | Epilepsy, progressive myoclonic 7, 616187 |
| KCNC3 | 76,9 | 64,2 | 94,7 | 88,4 | Spinocerebellar ataxia 13, 605259 |

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|---|---|---|---|---|---|
| KCNH1 | 98,7 | 98,5 | 98,7 | 98,7 | Zimmermann-Laband syndrome 1, 135500<br>Temple-Baraitser syndrome, 611816 |
| KCNJ10 | 89,2 | 88,5 | 100 | 100 | Enlarged vestibular aqueduct, digenic, 600791<br>SESAME syndrome, 612780 |
| KCNJ11 | 100 | 100 | 100 | 100 | Diabetes, permanent neonatal 2, with or without neurologic features, 618856<br>Maturity-onset diabetes of the young, type 13, 616329<br>Diabetes mellitus, transient neonatal 3, 610582<br>Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| KCNJ6 | 100 | 100 | 100 | 100 | Keppen-Lubinsky syndrome, 614098 |
| KCNK4 | 99,6 | 98,1 | 100 | 100 | Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 |
| KCNK9 | 97,3 | 97,3 | 97,3 | 97,3 | Birk-Barel syndrome, 612292 |
| KCNMA1 | 94 | 93 | 100 | 100 | Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446<br>Cerebellar atrophy, developmental delay, and seizures, 617643<br>Liang-Wang syndrome, 618729 |
| KCNN2 | 73,7 | 72,7 | 100 | 100 | No OMIM disease ID |
| KCNN3 | 100 | 99,7 | 100 | 100 | Zimmermann-Laband syndrome 3, 618658 |
| KCNQ2 | 91,2 | 89,1 | 100 | 100 | Developmental and epileptic encephalopathy 7, 613720<br>Seizures, benign neonatal, 1, 121200<br>Myokymia, 121200 |
| KCNQ3 | 99,8 | 97,2 | 99,5 | 98,7 | Seizures, benign neonatal, 2, 121201 |
| KCNQ5 | 97,4 | 95,3 | 100 | 100 | Mental retardation, autosomal dominant 46, 617601 |
| KCNT1 | 95,8 | 95 | 98,6 | 97,1 | Developmental and epileptic encephalopathy 14, 614959<br>Epilepsy nocturnal frontal lobe, 5, 615005 |
| KCNT2 | 99,1 | 96,9 | 100 | 99,9 | Developmental and epileptic encephalopathy 57, 617771 |
| KCTD3 | 99,7 | 99,4 | 100 | 100 | No OMIM disease ID |
| KCTD7 | 95 | 95 | 100 | 100 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 |
| KDM1A | 97,7 | 95,1 | 100 | 100 | Cleft palate, psychomotor retardation, and distinctive facial features, 616728 |
| KDM3B | 97,8 | 96,2 | 100 | 100 | Diets-Jongmans syndrome, 618846 |
| KDM4B | 99,8 | 98,6 | 100 | 100 | Intellectual developmental disorder, autosomal dominant 65, 619320 |
| KDM5B | 93,4 | 90,7 | 94,7 | 93,3 | Mental retardation, autosomal recessive 65, 618109 |
| KDM5C | 99,7 | 97,7 | 100 | 100 | Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 |
| KDM6A | 94,2 | 85,9 | 100 | 99,9 | Kabuki syndrome 2, 300867 |

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|---|---|---|---|---|---|
| KDM6B | 98,6 | 97,4 | 100 | 100 | Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505 |
| KIAA0586 | 97,1 | 92 | 95,8 | 95,7 | Short-rib thoracic dysplasia 14 with polydactyly, 616546<br>Joubert syndrome 23, 616490 |
| KIAA1109 | 99,8 | 99 | 100 | 100 | Alkuraya-Kucinskas syndrome, 617822 |
| KIDINS220 | 100 | 99,9 | 100 | 100 | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296<br>Ventriculomegaly and arthrogryposis, 619501 |
| KIF11 | 96,8 | 93,1 | 100 | 100 | Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 |
| KIF14 | 99,2 | 96,8 | 100 | 99,9 | Microcephaly 20, primary, autosomal recessive, 617914<br>?Meckel syndrome 12, 616258 |
| KIF1A | 97,4 | 95,3 | 98 | 98 | NESCAV syndrome, 614255<br>Neuropathy, hereditary sensory, type IIC, 614213<br>Spastic paraplegia 30, autosomal dominant, 610357<br>Spastic paraplegia 30, autosomal recessive, 610357 |
| KIF21B | 98,1 | 96,8 | 100 | 100 | No OMIM disease ID |
| KIF2A | 99 | 95,3 | 100 | 99,8 | Cortical dysplasia, complex, with other brain malformations 3, 615411 |
| KIF3B | 99,9 | 98,9 | 100 | 100 | Retinitis pigmentosa 89, 618955 |
| KIF4A | 98,4 | 92,2 | 100 | 100 | ?Intellectual developmental disorder, X-linked 100, 300923 |
| KIF5A | 100 | 99,8 | 100 | 100 | Myoclonus, intractable, neonatal, 617235<br>Spastic paraplegia 10, autosomal dominant, 604187 |
| KIF5C | 99,6 | 97 | 99,8 | 99,8 | Cortical dysplasia, complex, with other brain malformations 2, 615282 |
| KIF7 | 93,6 | 91,9 | 99,7 | 98,6 | Joubert syndrome 12, 200990<br>Acrocallosal syndrome, 200990<br>?Hydrolethalus syndrome 2, 614120<br>?Al-Gazali-Bakalinova syndrome, 607131 |
| KIFBP | 96,1 | 96 | 96,1 | 96,1 | Goldberg-Shprintzen megacolon syndrome, 609460 |
| KIRREL3 | 99,9 | 98,8 | 100 | 100 | No OMIM disease ID |
| KLF7 | 100 | 98,9 | 100 | 100 | No OMIM disease ID |
| KLHL15 | 99,9 | 99,1 | 100 | 100 | Intellectual developmental disorder, X-linked 103, 300982 |
| KMT2A | 100 | 99,7 | 100 | 99,8 | Wiedemann-Steiner syndrome, 605130 |
| KMT2B | 96,2 | 94 | 98,5 | 97,8 | Dystonia 28, childhood-onset, 617284 |
| KMT2C | 91,9 | 90,3 | 100 | 100 | Kleefstra syndrome 2, 617768 |
| KMT2D | 99,9 | 99 | 100 | 100 | Kabuki syndrome 1, 147920 |
| KMT2E | 99,6 | 98,1 | 100 | 100 | O'Donnell-Luria-Rodan syndrome, 618512 |

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|---|---|---|---|---|---|
| KMT5B | 99,7 | 98,5 | 100 | 100 | Mental retardation, autosomal dominant 51, 617788 |
| KNL1 | 99,1 | 97,3 | 98,9 | 98,7 | Microcephaly 4, primary, autosomal recessive, 604321 |
| KPTN | 100 | 100 | 100 | 100 | Mental retardation, autosomal recessive 41, 615637 |
| KRAS | 99 | 97,8 | 100 | 100 | Gastric cancer, somatic, 137215<br>Oculoectodermal syndrome, somatic, 600268<br>Breast cancer, somatic, 114480<br>Noonan syndrome 3, 609942<br>RAS-associated autoimmune leukoproliferative disorder, 614470<br>Arteriovenous malformation of the brain, somatic, 108010<br>Lung cancer, somatic, 211980<br>Pancreatic carcinoma, somatic, 260350<br>Leukemia, acute myeloid, somatic, 601626<br>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200<br>Cardiofaciocutaneous syndrome 2, 615278<br>Bladder cancer, somatic, 109800 |
| L1CAM | 100 | 98,8 | 100 | 100 | MASA syndrome, 303350<br>Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000<br>Corpus callosum, partial agenesis of, 304100<br>CRASH syndrome, 303350<br>Hydrocephalus with Hirschsprung disease, 307000<br>Hydrocephalus due to aqueductal stenosis, 307000 |
| L2HGDH | 98,9 | 96,4 | 100 | 100 | L-2-hydroxyglutaric aciduria, 236792 |
| LAMA1 | 99,9 | 99,3 | 100 | 100 | Poretti-Boltshauser syndrome, 615960 |
| LAMA2 | 99,9 | 99,1 | 100 | 100 | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138<br>Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 |
| LAMB1 | 100 | 99,6 | 100 | 100 | Lissencephaly 5, 615191 |
| LAMB2 | 99,9 | 99,3 | 100 | 100 | Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199<br>Pierson syndrome, 609049 |
| LAMC3 | 98,9 | 97,5 | 100 | 99,8 | Cortical malformations, occipital, 614115 |
| LAMP2 | 99,3 | 96 | 100 | 99,7 | Danon disease, 300257 |
| LARGE1 | 100 | 99,7 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 |
| LARP7 | 86,6 | 75,4 | 100 | 99,9 | Alazami syndrome, 615071 |
| LARS1 | 99,4 | 97,2 | 100 | 99,9 | ?Infantile liver failure syndrome 1, 615438 |
| LAS1L | 99,5 | 95,7 | 100 | 100 | Wilson-Turner syndrome, 309585 |

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|---|---|---|---|---|---|
| LIAS | 99,8 | 98,9 | 100 | 100 | Hyperglycinemia, lactic acidosis, and seizures, 614462 |
| LIG4 | 99,8 | 99,3 | 100 | 100 | LIG4 syndrome, 606593 |
| LINGO1 | 100 | 100 | 100 | 100 | Mental retardation, autosomal recessive 64, 618103 |
| LINS1 | 99,8 | 98,8 | 100 | 100 | Mental retardation, autosomal recessive 27, 614340 |
| LMAN2L | 100 | 99,5 | 100 | 100 | ?Mental retardation, autosomal recessive, 52, 616887 |
| LMBRD2 | 99,1 | 95,6 | 100 | 100 | No OMIM disease ID |
| LMNB1 | 99,9 | 99,2 | 100 | 100 | Leukodystrophy, adult-onset, autosomal dominant, 169500<br>Microcephaly 26, primary, autosomal dominant, 619179 |
| LMNB2 | 98,3 | 95,5 | 97,9 | 96,7 | Microcephaly 27, primary, autosomal dominant, 619180<br>?Epilepsy, progressive myoclonic, 9, 616540 |
| LONP1 | 100 | 99,9 | 100 | 100 | CODAS syndrome, 600373 |
| LRP2 | 100 | 99,8 | 100 | 100 | Donnai-Barrow syndrome, 222448 |
| LRPPRC | 99,7 | 99,3 | 100 | 99,9 | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 |
| LYRM7 | 95,6 | 86,5 | 100 | 99,2 | Mitochondrial complex III deficiency, nuclear type 8, 615838 |
| LYST | 99,4 | 97,8 | 100 | 100 | Chediak-Higashi syndrome, 214500 |
| LZTFL1 | 99,7 | 99,4 | 100 | 99,9 | Bardet-Biedl syndrome 17, 615994 |
| LZTR1 | 100 | 99,9 | 100 | 100 | Noonan syndrome 2, 605275<br>Noonan syndrome 10, 616564 |
| MAB21L1 | 100 | 100 | 100 | 100 | Cerebellar, ocular, craniofacial, and genital syndrome, 618479 |
| MAB21L2 | 100 | 100 | 100 | 100 | Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 |
| MACF1 | 99,6 | 98,7 | 100 | 100 | Lissencephaly 9 with complex brainstem malformation, 618325 |
| MADD | 100 | 99,2 | 100 | 100 | Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005<br>DEEAH syndrome, 619004 |
| MAF | 84,3 | 78,9 | 88,6 | 82,2 | Cataract 21, multiple types, 610202<br>Ayme-Gripp syndrome, 601088 |
| MAG | 100 | 100 | 100 | 100 | Spastic paraplegia 75, autosomal recessive, 616680 |
| MAGEL2 | 94,1 | 89,1 | 100 | 100 | Schaaf-Yang syndrome, 615547 |
| MAN1B1 | 100 | 99,7 | 100 | 100 | Rafiq syndrome, 614202 |
| MAN2B1 | 99,6 | 97,4 | 100 | 100 | Mannosidosis, alpha-, types I and II, 248500 |
| MANBA | 87,1 | 84,9 | 100 | 99,9 | Mannosidosis, beta, 248510 |
| MAOA | 100 | 99,8 | 99,4 | 97,9 | Brunner syndrome, 300615 |

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|---|---|---|---|---|---|
| MAP1B | 98,9 | 97,1 | 100 | 100 | Periventricular nodular heterotopia 9, 618918 |
| MAP2K1 | 99,6 | 96,1 | 100 | 100 | Cardiofaciocutaneous syndrome 3, 615279<br>Melorheostosis, isolated, somatic mosaic, 155950 |
| MAP2K2 | 98,5 | 95,3 | 100 | 100 | Cardiofaciocutaneous syndrome 4, 615280 |
| MAPK1 | 100 | 99,9 | 100 | 99,6 | Noonan syndrome 13, 619087 |
| MAPK8IP3 | 99,4 | 99 | 100 | 100 | Neurodevelopmental disorder with or without variable brain abnormalities, 618443 |
| MAPKAPK5 | 92,2 | 92,2 | 100 | 100 | No OMIM disease ID |
| MAPRE2 | 100 | 98,5 | 100 | 100 | Symmetric circumferential skin creases, congenital, 2, 616734 |
| MASP1 | 100 | 99,6 | 100 | 100 | 3MC syndrome 1, 257920 |
| MAST1 | 99,8 | 99,3 | 100 | 100 | Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 |
| MAT1A | 99,9 | 98,5 | 100 | 100 | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850<br>Methionine adenosyltransferase deficiency, autosomal recessive, 250850 |
| MBD5 | 99,9 | 99,8 | 100 | 100 | Mental retardation, autosomal dominant 1, 156200 |
| MBOAT7 | 100 | 99,3 | 100 | 100 | Mental retardation, autosomal recessive 57, 617188 |
| MBTPS2 | 99,9 | 98,5 | 100 | 100 | Keratosis follicularis spinulosa decalvans, X-linked, 308800<br>Osteogenesis imperfecta, type XIX, 301014<br>IFAP syndrome with or without BRESHECK syndrome, 308205<br>?Olmsted syndrome, X-linked, 300918 |
| MCCC1 | 99,9 | 98,7 | 100 | 100 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 |
| MCCC2 | 99,9 | 99,1 | 100 | 100 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 |
| MCOLN1 | 99,8 | 98,8 | 100 | 100 | Mucolipidosis IV, 252650 |
| MCPH1 | 99,8 | 98,6 | 100 | 100 | Microcephaly 1, primary, autosomal recessive, 251200 |
| MDH2 | 98 | 98 | 100 | 100 | Developmental and epileptic encephalopathy 51, 617339 |
| MECP2 | 99,8 | 97,5 | 100 | 99,7 | Intellectual developmental disorder, X-linked, syndromic 13, 300055<br>Rett syndrome, atypical, 312750<br>Encephalopathy, neonatal severe, 300673<br>Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260<br>Rett syndrome, 312750<br>Rett syndrome, preserved speech variant, 312750 |
| MECR | 100 | 98,7 | 100 | 100 | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 |

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|---|---|---|---|---|---|
| MED12 | 99,3 | 94,1 | 100 | 100 | Lujan-Fryns syndrome, 309520<br>Ohdo syndrome, X-linked, 300895<br>Opitz-Kaveggia syndrome, 305450 |
| MED13 | 99,9 | 99,6 | 100 | 100 | Intellectual developmental disorder 61, 618009 |
| MED13L | 100 | 99,5 | 100 | 100 | Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789<br>Transposition of the great arteries, dextro-looped 1, 608808 |
| MED17 | 95,8 | 92,4 | 100 | 100 | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 |
| MED23 | 99,9 | 99 | 100 | 100 | Mental retardation, autosomal recessive 18, 614249 |
| MED25 | 100 | 99,9 | 100 | 99,9 | Basel-Vanagait-Smirin-Yosef syndrome, 616449 |
| MED27 | 79 | 65,9 | 84,7 | 84,7 | Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 |
| MEF2C | 99,7 | 95,7 | 100 | 100 | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 |
| MEGF8 | 100 | 99,2 | 100 | 100 | Carpenter syndrome 2, 614976 |
| MEIS2 | 100 | 99,7 | 100 | 100 | Cleft palate, cardiac defects, and mental retardation, 600987 |
| METTL23 | 100 | 100 | 100 | 100 | Mental retardation, autosomal recessive 44, 615942 |
| METTL5 | 98,9 | 97,4 | 99,8 | 97,6 | Intellectual developmental disorder, autosomal recessive 72, 618665 |
| MFF | 93,9 | 89,4 | 100 | 100 | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 |
| MFSD2A | 99,5 | 97,3 | 100 | 100 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 |
| MFSD8 | 99,6 | 99,4 | 100 | 100 | Macular dystrophy with central cone involvement, 616170<br>Ceroid lipofuscinosis, neuronal, 7, 610951 |
| MGAT2 | 100 | 99,9 | 100 | 100 | Congenital disorder of glycosylation, type IIa, 212066 |
| MGP | 98,7 | 93,6 | 100 | 100 | Keutel syndrome, 245150 |
| MIA3 | 99,8 | 99,3 | 100 | 100 | ?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269 |
| MICU1 | 97,3 | 92,2 | 100 | 100 | Myopathy with extrapyramidal signs, 615673 |
| MID1 | 99,6 | 97,7 | 100 | 100 | Opitz GBBB syndrome, type I, 300000 |
| MID2 | 99,8 | 98,6 | 99,9 | 99,8 | ?Intellectual developmental disorder, X-linked 101, 300928 |
| MINPP1 | 99,7 | 99,3 | 100 | 99,9 | No OMIM disease ID |
| MKKS | 100 | 100 | 100 | 100 | McKusick-Kaufman syndrome, 236700<br>Bardet-Biedl syndrome 6, 605231 |
| MKS1 | 99,4 | 96,3 | 100 | 100 | Bardet-Biedl syndrome 13, 615990<br>Meckel syndrome 1, 249000<br>Joubert syndrome 28, 617121 |

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|---|---|---|---|---|---|
| MLC1 | 100 | 98,8 | 100 | 100 | Megalencephalic leukoencephalopathy with subcortical cysts, 604004 |
| MLYCD | 96,8 | 92,5 | 100 | 99,4 | Malonyl-CoA decarboxylase deficiency, 248360 |
| MMAA | 100 | 100 | 100 | 100 | Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 |
| MMAB | 100 | 99,9 | 100 | 100 | Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 |
| MMACHC | 100 | 100 | 100 | 100 | Methylmalonic aciduria and homocystinuria, cblC type, 277400 |
| MMADHC | 91,6 | 81,3 | 89,7 | 89,7 | Methylmalonic aciduria, cblD type, variant 2, 277410<br>Methylmalonic aciduria and homocystinuria, cblD type, 277410<br>Homocystinuria, cblD type, variant 1, 277410 |
| MMGT1 | 99,2 | 98,4 | 100 | 99,8 | No OMIM disease ID |
| MMUT | 99,7 | 98,2 | 100 | 100 | Methylmalonic aciduria, mut(0) type, 251000 |
| MN1 | 100 | 99,7 | 100 | 100 | CEBALID syndrome, 618774<br>Meningioma, 607174 |
| MOCS1 | 98,9 | 95,5 | 100 | 100 | Molybdenum cofactor deficiency A, 252150 |
| MOCS2 | 99,4 | 99,4 | 100 | 100 | Molybdenum cofactor deficiency B, 252160 |
| MOGS | 100 | 99,9 | 100 | 100 | Congenital disorder of glycosylation, type IIb, 606056 |
| MORC2 | 100 | 99,5 | 100 | 100 | Charcot-Marie-Tooth disease, axonal, type 2Z, 616688<br>Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 |
| MPDU1 | 100 | 99,2 | 100 | 100 | Congenital disorder of glycosylation, type If, 609180 |
| MPDZ | 99,8 | 98,5 | 100 | 100 | Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 |
| MPLKIP | 100 | 99,4 | 100 | 100 | Trichothiodystrophy 4, nonphotosensitive, 234050 |
| MPV17 | 100 | 98,7 | 100 | 100 | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400<br>Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 |
| MRAS | 100 | 99,3 | 100 | 100 | Noonan syndrome 11, 618499 |
| MRPS22 | 99,7 | 98,3 | 100 | 100 | Ovarian dysgenesis 7, 618117<br>Combined oxidative phosphorylation deficiency 5, 611719 |
| MSL2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| MSL3 | 83,4 | 75,3 | 97,8 | 96,6 | Basilicata-Akhtar syndrome, 301032 |
| MSMO1 | 93,1 | 86,8 | 100 | 100 | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 |
| MTFMT | 99,9 | 99,5 | 100 | 100 | Combined oxidative phosphorylation deficiency 15, 614947<br>Mitochondrial complex I deficiency, nuclear type 27, 618248 |
| MTHFR | 97,3 | 95,9 | 100 | 100 | Homocystinuria due to MTHFR deficiency, 236250 |
| MTHFS | 75 | 75 | 100 | 100 | Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 |

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|---|---|---|---|---|---|
| MTO1 | 90,9 | 88,8 | 92,8 | 91,4 | Combined oxidative phosphorylation deficiency 10, 614702 |
| MTOR | 99,9 | 98,9 | 100 | 100 | Focal cortical dysplasia, type II, somatic, 607341<br>Smith-Kingsmore syndrome, 616638 |
| MTR | 100 | 99,9 | 100 | 100 | Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 |
| C12orf65 | 99 | 94,5 | 100 | 100 | Spastic paraplegia 55, autosomal recessive, 615035<br>Combined oxidative phosphorylation deficiency 7, 613559 |
| MTRR | 99,8 | 98,4 | 100 | 100 | Homocystinuria-megaloblastic anemia, cbl E type, 236270 |
| MVK | 91,4 | 90,5 | 90,5 | 90,5 | Hyper-IgD syndrome, 260920<br>Porokeratosis 3, multiple types, 175900<br>Mevalonic aciduria, 610377 |
| MYCN | 100 | 100 | 99,2 | 96,2 | Feingold syndrome 1, 164280 |
| MYH9 | 99,9 | 98,9 | 100 | 100 | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100<br>Deafness, autosomal dominant 17, 603622 |
| MYO5A | 99,6 | 98,3 | 100 | 100 | Griscelli syndrome, type 1, 214450 |
| MYO9A | 99,8 | 98,9 | 100 | 100 | Myasthenic syndrome, congenital, 24, presynaptic, 618198 |
| MYT1L | 87 | 86,3 | 90,2 | 90,1 | Mental retardation, autosomal dominant 39, 616521 |
| NAA10 | 99,8 | 97,9 | 99,9 | 99,9 | Microphthalmia, syndromic 1, 309800<br>Ogden syndrome, 300855 |
| NAA15 | 94,8 | 91,2 | 96,8 | 96,7 | Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 |
| NAA20 | 99,9 | 99,3 | 100 | 100 | No OMIM disease ID |
| NACC1 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 |
| NAGA | 100 | 100 | 100 | 100 | Schindler disease, type I, 609241<br>Kanzaki disease, 609242<br>Schindler disease, type III, 609241 |
| NAGLU | 93,8 | 91,7 | 99,9 | 98,7 | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491<br>Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 |
| NALCN | 99,7 | 98,9 | 99,8 | 99,7 | Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266<br>Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 |
| NANS | 100 | 99,9 | 100 | 100 | Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 |
| NARS1 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092<br>Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 |

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|---|---|---|---|---|---|
| NARS2 | 97,9 | 97,1 | 100 | 99,9 | Combined oxidative phosphorylation deficiency 24, 616239<br>?Deafness, autosomal recessive 94, 618434 |
| NAXE | 100 | 98,6 | 100 | 100 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 |
| NBEA | 91,8 | 90,3 | 100 | 100 | Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 |
| NBN | 99,2 | 97,8 | 100 | 99,9 | Leukemia, acute lymphoblastic, 613065<br>Aplastic anemia, 609135<br>Nijmegen breakage syndrome, 251260 |
| NCAPG2 | 99,8 | 99 | 100 | 100 | Khan-Khan-Katsanis syndrome, 618460 |
| NCDN | 100 | 99,9 | 100 | 100 | Neurodevelopmental disorder with infantile epileptic spasms, 619373 |
| NCKAP1 | 98,5 | 96,1 | 100 | 100 | No OMIM disease ID |
| NDE1 | 100 | 99,4 | 100 | 100 | Lissencephaly 4 (with microcephaly), 614019<br>?Microhydranencephaly, 605013 |
| NDP | 100 | 99,7 | 100 | 100 | Exudative vitreoretinopathy 2, X-linked, 305390<br>Norrie disease, 310600 |
| NDST1 | 100 | 100 | 100 | 100 | Mental retardation, autosomal recessive 46, 616116 |
| NDUFA1 | 99,8 | 99,3 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 12, 301020 |
| NDUFA11 | 100 | 99,8 | 100 | 99,9 | Mitochondrial complex I deficiency, nuclear type 14, 618236 |
| NDUFA12 | 99,6 | 99,6 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 23, 618244 |
| NDUFA2 | 100 | 100 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 13, 618235 |
| NDUFA8 | 100 | 97,3 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 37, 619272 |
| NDUFAF3 | 100 | 99,9 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 18, 618240 |
| NDUFAF5 | 99,7 | 99,1 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 16, 618238 |
| NDUFAF8 | 62,6 | 61,7 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 34, 618776 |
| NDUFS1 | 99,9 | 99,1 | 100 | 99,9 | Mitochondrial complex I deficiency, nuclear type 5, 618226 |
| NDUFS2 | 100 | 100 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 6, 618228 |
| NDUFS3 | 90,7 | 90,6 | 92,8 | 90,7 | Mitochondrial complex I deficiency, nuclear type 8, 618230 |
| NDUFS4 | 99,7 | 99,7 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 1, 252010 |
| NDUFS6 | 100 | 99,8 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 9, 618232 |
| NDUFS7 | 100 | 99,7 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 3, 618224 |
| NDUFS8 | 100 | 99,1 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 2, 618222 |
| NDUFV1 | 99 | 97 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 4, 618225 |

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|---|---|---|---|---|---|
| NDUFV2 | 85,8 | 78,7 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 7, 618229 |
| NEDD4L | 71,9 | 71,7 | 100 | 99,9 | Periventricular nodular heterotopia 7, 617201 |
| NEMF | 99,6 | 98,3 | 100 | 99,9 | Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 |
| NEU1 | 99,3 | 96,1 | 100 | 100 | Sialidosis, type II, 256550<br>Sialidosis, type I, 256550 |
| NEUROD2 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 72, 618374 |
| NEXMIF | 99,9 | 99 | 100 | 100 | Intellectual developmental disorder, X-linked 98, 300912 |
| NF1 | 91,8 | 89,3 | 100 | 100 | Watson syndrome, 193520<br>Leukemia, juvenile myelomonocytic, 607785<br>Neurofibromatosis, familial spinal, 162210<br>Neurofibromatosis, type 1, 162200<br>Neurofibromatosis-Noonan syndrome, 601321 |
| NFE2L2 | 100 | 99,9 | 100 | 100 | Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 |
| NFIA | 99,2 | 98,4 | 99,2 | 99,2 | Brain malformations with or without urinary tract defects, 613735 |
| NFIB | 97,4 | 96 | 100 | 99,9 | Macrocephaly, acquired, with impaired intellectual development, 618286 |
| NFIX | 100 | 99,3 | 99,4 | 98,6 | Marshall-Smith syndrome, 602535<br>Sotos syndrome 2, 614753 |
| NFU1 | 98,7 | 87,7 | 100 | 100 | Multiple mitochondrial dysfunctions syndrome 1, 605711 |
| NGLY1 | 99,8 | 99,7 | 100 | 100 | Congenital disorder of deglycosylation, 615273 |
| NHS | 96,1 | 94,1 | 100 | 100 | Cataract 40, X-linked, 302200<br>Nance-Horan syndrome, 302350 |
| NIPBL | 98,4 | 96,3 | 100 | 99,9 | Cornelia de Lange syndrome 1, 122470 |
| NKAP | 99 | 94,1 | 99,9 | 99,4 | Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039 |
| NKX2-1 | 99,3 | 89,3 | 100 | 100 | Chorea, hereditary benign, 118700<br>Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 |
| NLGN2 | 93,7 | 88,6 | 100 | 100 | No OMIM disease ID |
| NLGN3 | 99,7 | 98,2 | 100 | 100 | No OMIM disease ID |
| NLGN4X | 99,8 | 98,2 | 100 | 100 | Intellectual developmental disorder, X-linked, 300495 |
| NONO | 99,8 | 97,2 | 100 | 100 | Intellectual developmental disorder, X-linked syndromic 34, 300967 |
| NOVA2 | 99 | 92,8 | 96,8 | 92,8 | Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 |
| NPC1 | 99,9 | 99 | 100 | 100 | Niemann-Pick disease, type C1, 257220<br>Niemann-Pick disease, type D, 257220 |

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|---|---|---|---|---|---|
| NPC2 | 100 | 99,2 | 100 | 100 | Niemann-pick disease, type C2, 607625 |
| NPHP1 | 99,8 | 99,1 | 100 | 100 | Joubert syndrome 4, 609583<br>Nephronophthisis 1, juvenile, 256100<br>Senior-Loken syndrome-1, 266900 |
| NR2F1 | 100 | 100 | 98,2 | 93 | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 |
| NR4A2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| NRAS | 100 | 100 | 100 | 100 | Noonan syndrome 6, 613224<br>?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470<br>Melanocytic nevus syndrome, congenital, somatic, 137550<br>Epidermal nevus, somatic, 162900<br>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200<br>Thyroid carcinoma, follicular, somatic, 188470<br>Neurocutaneous melanosis, somatic, 249400<br>Colorectal cancer, somatic, 114500 |
| NRROS | 100 | 100 | 100 | 100 | Seizures, early-onset, with neurodegeneration and brain calcification, 618875 |
| NRXN1 | 97,5 | 96,9 | 99,9 | 99,7 | Pitt-Hopkins-like syndrome 2, 614325 |
| NSD1 | 100 | 99,8 | 100 | 100 | Sotos syndrome 1, 117550 |
| NSD2 | 99,9 | 98,3 | 100 | 100 | No OMIM disease ID |
| NSDHL | 99,8 | 96,3 | 100 | 100 | CK syndrome, 300831<br>CHILD syndrome, 308050 |
| NSF | 99,5 | 99,3 | 100 | 100 | Developmental and epileptic encephalopathy 96, 619340 |
| NSUN2 | 95,5 | 92,9 | 100 | 100 | Mental retardation, autosomal recessive 5, 611091 |
| NT5C2 | 97,7 | 94,6 | 100 | 100 | Spastic paraplegia 45, autosomal recessive, 613162 |
| NTNG2 | 99,1 | 97,3 | 100 | 99,6 | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 |
| NTRK1 | 99,9 | 98,5 | 100 | 100 | Insensitivity to pain, congenital, with anhidrosis, 256800 |
| NTRK2 | 100 | 99,9 | 100 | 100 | Developmental and epileptic encephalopathy 58, 617830<br>Obesity, hyperphagia, and developmental delay, 613886 |
| NUBPL | 99,5 | 96,9 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 21, 618242 |
| NUDT2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| NUP107 | 99,7 | 98,4 | 100 | 99,9 | ?Ovarian dysgenesis 6, 618078<br>Galloway-Mowat syndrome 7, 618348<br>Nephrotic syndrome, type 11, 616730 |
| NUP188 | 99,9 | 99,1 | 100 | 100 | Sandestig-Stefanova syndrome, 618804 |

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| NUP214 | 99,8 | 99 | 100 | 100 | Leukemia, T-cell acute lymphoblastic, somatic, 613065<br>Leukemia, acute myeloid, somatic, 601626 |
| NUP62 | 100 | 100 | 100 | 100 | Striatonigral degeneration, infantile, 271930 |
| NUS1 | 56,5 | 42 | 100 | 99,9 | Mental retardation, autosomal dominant 55, with seizures, 617831<br>?Congenital disorder of glycosylation, type 1aa, 617082 |
| OAT | 82 | 73 | 100 | 100 | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 |
| OCLN | 100 | 99,9 | 100 | 100 | Pseudo-TORCH syndrome 1, 251290 |
| OCRL | 99,4 | 97,6 | 100 | 99,9 | Dent disease 2, 300555<br>Lowe syndrome, 309000 |
| ODC1 | 100 | 99,3 | 100 | 100 | Bachmann-Bupp syndrome, 619075 |
| OFD1 | 87,1 | 71,3 | 100 | 99,8 | Simpson-Golabi-Behmel syndrome, type 2, 300209<br>?Retinitis pigmentosa 23, 300424<br>Orofaciodigital syndrome I, 311200<br>Joubert syndrome 10, 300804 |
| OGT | 99,7 | 98,3 | 100 | 100 | Intellectual developmental disorder, X-linked 106, 300997 |
| OPA3 | 100 | 99,5 | 100 | 100 | 3-methylglutaconic aciduria, type III, 258501<br>Optic atrophy 3 with cataract, 165300 |
| OPHN1 | 99,3 | 96,3 | 99,5 | 98,4 | Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 |
| ORC1 | 99,9 | 97,9 | 100 | 100 | Meier-Gorlin syndrome 1, 224690 |
| OSGEP | 99,8 | 95,7 | 100 | 100 | Galloway-Mowat syndrome 3, 617729 |
| OTC | 100 | 99,9 | 100 | 99,7 | Ornithine transcarbamylase deficiency, 311250 |
| OTUD5 | 89 | 76,4 | 98,4 | 95,2 | Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 |
| OTUD6B | 99,7 | 98,6 | 100 | 99,8 | Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 |
| OTX2 | 100 | 99 | 100 | 100 | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125<br>Pituitary hormone deficiency, combined, 6, 613986<br>Microphthalmia, syndromic 5, 610125 |
| OXR1 | 99,2 | 96,3 | 100 | 99,9 | Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 |
| P4HTM | 99,3 | 97,6 | 100 | 99,6 | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 |
| PACS1 | 99,8 | 99 | 100 | 100 | Schuurs-Hoeijmakers syndrome, 615009 |
| PACS2 | 99,8 | 97,1 | 99,9 | 99,6 | Developmental and epileptic encephalopathy 66, 618067 |
| PAFAH1B1 | 93,5 | 84,9 | 100 | 100 | Subcortical laminar heterotopia, 607432<br>Lissencephaly 1, 607432 |

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|---|---|---|---|---|---|
| PAH | 100 | 100 | 100 | 100 | Phenylketonuria, 261600 |
| PAK1 | 100 | 99,4 | 100 | 100 | Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 |
| PAK3 | 98,9 | 93,6 | 100 | 99,5 | Intellectual developmental disorder, X-linked 30, 300558 |
| MPP5 | 99,8 | 99,1 | 100 | 100 | No OMIM disease ID |
| PAM16 | 65,3 | 65,2 | 82,9 | 82,9 | Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 |
| PANK2 | 100 | 99,7 | 100 | 100 | HARP syndrome, 607236<br>Neurodegeneration with brain iron accumulation 1, 234200 |
| PANX1 | 100 | 100 | 100 | 100 | Oocyte maturation defect 7, 618550 |
| PARN | 81,1 | 80,4 | 88,3 | 87,6 | Dyskeratosis congenita, autosomal recessive 6, 616353<br>Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 |
| PAX1 | 92,6 | 87,5 | 100 | 99,7 | Otofaciocervical syndrome 2, 615560 |
| PAX6 | 100 | 99,9 | 100 | 100 | Optic nerve hypoplasia, 165550<br>Cataract with late-onset corneal dystrophy, 106210<br>?Coloboma, ocular, 120200<br>?Coloboma of optic nerve, 120430<br>Aniridia, 106210<br>Anterior segment dysgenesis 5, multiple subtypes, 604229<br>?Morning glory disc anomaly, 120430<br>Foveal hypoplasia 1, 136520<br>Keratitis, 148190 |
| PAX7 | 100 | 100 | 100 | 100 | Rhabdomyosarcoma 2, alveolar, 268220<br>Myopathy, congenital, progressive, with scoliosis, 618578 |
| PAX8 | 100 | 99,6 | 100 | 100 | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 |
| PBX1 | 100 | 99,1 | 100 | 100 | Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 |
| PC | 99,7 | 98 | 100 | 100 | Pyruvate carboxylase deficiency, 266150 |
| PCCA | 98,9 | 93,4 | 100 | 100 | Propionicacidemia, 606054 |
| PCCB | 96,7 | 95,4 | 99 | 96,2 | Propionicacidemia, 606054 |
| PCDH12 | 100 | 100 | 100 | 100 | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 |
| PCDH19 | 99,7 | 97,7 | 100 | 100 | Developmental and epileptic encephalopathy 9, 300088 |
| PCDHGC4 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PCGF2 | 99,6 | 93,3 | 100 | 100 | Turnpenny-Fry syndrome, 618371 |
| PCLO | 99,4 | 98,3 | 100 | 100 | ?Pontocerebellar hypoplasia, type 3, 608027 |

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|---|---|---|---|---|---|
| PCNT | 99,3 | 96,5 | 100 | 100 | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 |
| PCYT2 | 100 | 98,3 | 99,6 | 97,8 | Spastic paraplegia 82, autosomal recessive, 618770 |
| PDE2A | 100 | 99,5 | 100 | 100 | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 |
| PDE4D | 95,7 | 93,1 | 100 | 99,6 | Acrodysostosis 2, with or without hormone resistance, 614613 |
| PDGFRB | 99,2 | 97,3 | 100 | 100 | Premature aging syndrome, Penttinen type, 601812<br>Kosaki overgrowth syndrome, 616592<br>Myofibromatosis, infantile, 1, 228550<br>Basal ganglia calcification, idiopathic, 4, 615007 |
| PDHA1 | 98,8 | 95,9 | 100 | 100 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 |
| PDHB | 99,2 | 96,8 | 100 | 100 | Pyruvate dehydrogenase E1-beta deficiency, 614111 |
| PDHX | 99,8 | 99,6 | 100 | 100 | Lacticacidemia due to PDX1 deficiency, 245349 |
| PDP1 | 100 | 100 | 100 | 100 | Pyruvate dehydrogenase phosphatase deficiency, 608782 |
| PDSS1 | 95,2 | 87,8 | 97,4 | 97,4 | Coenzyme Q10 deficiency, primary, 2, 614651 |
| PDSS2 | 98,4 | 94,3 | 100 | 100 | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PEPD | 100 | 99,4 | 100 | 100 | Prolidase deficiency, 170100 |
| PET100 | 100 | 99,2 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 12, 619055 |
| PEX1 | 99,8 | 99,4 | 100 | 100 | Heimler syndrome 1, 234580<br>Peroxisome biogenesis disorder 1B (NALD/IRD), 601539<br>Peroxisome biogenesis disorder 1A (Zellweger), 214100 |
| PEX10 | 98,8 | 90,6 | 100 | 100 | Peroxisome biogenesis disorder 6A (Zellweger), 614870<br>Peroxisome biogenesis disorder 6B, 614871 |
| PEX11B | 100 | 98,3 | 100 | 100 | Peroxisome biogenesis disorder 14B, 614920 |
| PEX12 | 100 | 100 | 100 | 100 | Peroxisome biogenesis disorder 3B, 266510<br>Peroxisome biogenesis disorder 3A (Zellweger), 614859 |
| PEX13 | 100 | 100 | 100 | 100 | Peroxisome biogenesis disorder 11A (Zellweger), 614883<br>Peroxisome biogenesis disorder 11B, 614885 |
| PEX16 | 97,1 | 93,9 | 100 | 100 | Peroxisome biogenesis disorder 8B, 614877<br>Peroxisome biogenesis disorder 8A (Zellweger), 614876 |
| PEX19 | 99 | 94,4 | 100 | 100 | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| PEX2 | 100 | 100 | 100 | 100 | Peroxisome biogenesis disorder 5A (Zellweger), 614866<br>Peroxisome biogenesis disorder 5B, 614867 |
| PEX26 | 100 | 99,8 | 100 | 100 | Peroxisome biogenesis disorder 7B, 614873<br>Peroxisome biogenesis disorder 7A (Zellweger), 614872 |

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| PEX3 | 99,4 | 99,2 | 100 | 100 | Peroxisome biogenesis disorder 10A (Zellweger), 614882<br>?Peroxisome biogenesis disorder 10B, 617370 |
| PEX5 | 99,9 | 98,8 | 100 | 100 | Peroxisome biogenesis disorder 2B, 202370<br>Peroxisome biogenesis disorder 2A (Zellweger), 214110<br>Rhizomelic chondrodysplasia punctata, type 5, 616716 |
| PEX6 | 96,4 | 88 | 100 | 100 | Peroxisome biogenesis disorder 4B, 614863<br>Peroxisome biogenesis disorder 4A (Zellweger), 614862<br>Heimler syndrome 2, 616617 |
| PEX7 | 88 | 81 | 91,3 | 91,2 | Rhizomelic chondrodysplasia punctata, type 1, 215100<br>Peroxisome biogenesis disorder 9B, 614879 |
| PGAP1 | 98,7 | 94,6 | 100 | 99,8 | Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 |
| PGAP2 | 100 | 99,9 | 100 | 100 | Hyperphosphatasia with mental retardation syndrome 3, 614207 |
| PGAP3 | 62,6 | 58,1 | 100 | 100 | Hyperphosphatasia with mental retardation syndrome 4, 615716 |
| PGK1 | 90,3 | 73,2 | 100 | 100 | Phosphoglycerate kinase 1 deficiency, 300653 |
| PGM2L1 | 99,8 | 97,7 | 100 | 100 | No OMIM disease ID |
| PGM3 | 99,9 | 99,7 | 91,7 | 91,7 | Immunodeficiency 23, 615816 |
| PHACTR1 | 100 | 99,6 | 100 | 99,9 | Developmental and epileptic encephalopathy 70, 618298 |
| PHF21A | 100 | 99,7 | 100 | 100 | Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 |
| PHF6 | 96,2 | 84,7 | 100 | 99,2 | Borjeson-Forssman-Lehmann syndrome, 301900 |
| PHF8 | 98,9 | 94,4 | 100 | 100 | Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263 |
| PHGDH | 99,9 | 98,2 | 100 | 100 | Neu-Laxova syndrome 1, 256520<br>Phosphoglycerate dehydrogenase deficiency, 601815 |
| PHIP | 98,2 | 95,7 | 99,9 | 99,6 | Chung-Jansen syndrome, 617991 |
| PI4KA | 92,6 | 88,7 | 100 | 99,9 | Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 |
| PIBF1 | 99,1 | 95 | 100 | 99,9 | Joubert syndrome 33, 617767 |
| PIDD1 | 100 | 99,5 | 100 | 100 | No OMIM disease ID |
| PIGA | 91,6 | 82,5 | 100 | 99,8 | Paroxysmal nocturnal hemoglobinuria, somatic, 300818<br>Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 |
| PIGB | 99,5 | 97,3 | 100 | 100 | Developmental and epileptic encephalopathy 80, 618580 |
| PIGC | 96 | 86,2 | 100 | 100 | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 |
| PIGG | 100 | 99,6 | 100 | 100 | Mental retardation, autosomal recessive 53, 616917 |
| PIGH | 81,9 | 64,4 | 75,9 | 74,4 | Glycosylphosphatidylinositol biosynthesis defect 17, 618010 |

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|---|---|---|---|---|---|
| PIGK | 98,8 | 94,2 | 100 | 100 | Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 |
| PIGL | 100 | 99,6 | 100 | 100 | CHIME syndrome, 280000 |
| PIGN | 93,1 | 89,6 | 98,8 | 98,6 | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGO | 100 | 99,8 | 100 | 100 | Hyperphosphatasia with mental retardation syndrome 2, 614749 |
| PIGP | 95,6 | 85,5 | 100 | 99,9 | Developmental and epileptic encephalopathy 55, 617599 |
| PIGQ | 93,4 | 91,6 | 100 | 100 | Developmental and epileptic encephalopathy 77, 618548 |
| PIGS | 100 | 99,6 | 100 | 100 | Developmental and epileptic encephalopathy 95, 618143 |
| PIGT | 98,1 | 98 | 100 | 100 | ?Paroxysmal nocturnal hemoglobinuria 2, 615399<br>Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 |
| PIGU | 100 | 99,5 | 100 | 98,9 | Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 |
| PIGV | 100 | 100 | 100 | 100 | Hyperphosphatasia with mental retardation syndrome 1, 239300 |
| PIGW | 100 | 99,7 | 100 | 100 | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 |
| PIGY | 100 | 100 | 100 | 100 | Hyperphosphatasia with mental retardation syndrome 6, 616809 |
| PIK3CA | 97,7 | 97,3 | 100 | 100 | CLOVE syndrome, somatic, 612918<br>Hepatocellular carcinoma, somatic, 114550<br>Breast cancer, somatic, 114480<br>Ovarian cancer, somatic, 167000<br>Colorectal cancer, somatic, 114500<br>Macrodactyly, somatic, 155500<br>CLAPO syndrome, somatic, 613089<br>Keratosis, seborrheic, somatic, 182000<br>Nevus, epidermal, somatic, 162900<br>Gastric cancer, somatic, 613659<br>Nonsmall cell lung cancer, somatic, 211980<br>Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501<br>Cowden syndrome 5, 615108 |
| PIK3R2 | 90,9 | 89,1 | 99,7 | 98 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 |
| PISD | 100 | 99,7 | 100 | 100 | Liberfarb syndrome, 618889 |
| PITRM1 | 98,2 | 96,2 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 30, 619405 |
| PJA1 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PLA2G6 | 92,1 | 90,7 | 92,3 | 92,3 | Parkinson disease 14, autosomal recessive, 612953<br>Neurodegeneration with brain iron accumulation 2B, 610217<br>Infantile neuroaxonal dystrophy 1, 256600 |

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|---|---|---|---|---|---|
| PLAA | 99,6 | 98,4 | 100 | 100 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 |
| PLCB1 | 99,9 | 99,4 | 100 | 100 | Developmental and epileptic encephalopathy 12, 613722 |
| PLK1 | 100 | 99 | 100 | 100 | No OMIM disease ID |
| PLK4 | 99,4 | 98,4 | 100 | 100 | Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 |
| PLP1 | 99,9 | 97,7 | 100 | 100 | Pelizaeus-Merzbacher disease, 312080<br>Spastic paraplegia 2, X-linked, 312920 |
| PLPBP | 95,1 | 88,9 | 100 | 99,9 | Epilepsy, early-onset, vitamin B6-dependent, 617290 |
| PLXNA1 | 100 | 99,9 | 100 | 100 | No OMIM disease ID |
| PLXND1 | 98,9 | 96,1 | 99,8 | 99,5 | No OMIM disease ID |
| PMM2 | 99,8 | 99,8 | 100 | 100 | Congenital disorder of glycosylation, type Ia, 212065 |
| PMPCA | 97,6 | 93,5 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 2, 213200 |
| PMPCB | 99,9 | 99,2 | 100 | 100 | Multiple mitochondrial dysfunctions syndrome 6, 617954 |
| PNKP | 100 | 100 | 100 | 100 | ?Charcot-Marie-Tooth disease, type 2B2, 605589<br>Ataxia-oculomotor apraxia 4, 616267<br>Microcephaly, seizures, and developmental delay, 613402 |
| PNP | 99,8 | 98,7 | 100 | 100 | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 |
| PNPLA6 | 100 | 99,8 | 100 | 100 | Spastic paraplegia 39, autosomal recessive, 612020<br>Oliver-McFarlane syndrome, 275400<br>?Laurence-Moon syndrome, 245800<br>Boucher-Neuhauser syndrome, 215470 |
| POGZ | 99,4 | 98,8 | 100 | 100 | White-Sutton syndrome, 616364 |
| POLA1 | 99 | 93,6 | 100 | 99,7 | Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220<br>Van Esch-O'Driscoll syndrome, 301030 |
| POLG | 99,9 | 98,8 | 100 | 100 | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459<br>Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662<br>Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700<br>Progressive external ophthalmoplegia, autosomal dominant 1, 157640<br>Progressive external ophthalmoplegia, autosomal recessive 1, 258450 |
| POLR1C | 89,6 | 84,8 | 82,8 | 82,8 | Leukodystrophy, hypomyelinating, 11, 616494<br>Treacher Collins syndrome 3, 248390 |
| POLR2A | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 |

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|---|---|---|---|---|---|
| POLR3A | 99,9 | 99 | 100 | 100 | Wiedemann-Rautenstrauch syndrome, 264090<br>Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 |
| POLR3B | 99,7 | 97,6 | 100 | 100 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 |
| POMGNT1 | 100 | 99,8 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151<br>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157<br>Retinitis pigmentosa 76, 617123<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 |
| POMGNT2 | 100 | 100 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 |
| POMK | 100 | 100 | 100 | 100 | ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 |
| POMT1 | 99,5 | 97,3 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670<br>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 |
| POMT2 | 99,8 | 97,3 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 |
| PORCN | 100 | 99,1 | 100 | 100 | Focal dermal hypoplasia, 305600 |
| POU1F1 | 99,9 | 98,2 | 100 | 100 | Pituitary hormone deficiency, combined, 1, 613038 |
| POU3F3 | 76,5 | 63,8 | 90,8 | 79,1 | Snijders Blok-Fisher syndrome, 618604 |
| PPIL1 | 100 | 100 | 100 | 100 | Pontocerebellar hypoplasia, type 14, 619301 |
| PPM1D | 100 | 99,9 | 100 | 100 | Breast cancer, somatic, 114480<br>Jansen de Vries syndrome, 617450 |
| PPP1CB | 99,8 | 98,7 | 100 | 100 | Noonan syndrome-like disorder with loose anagen hair 2, 617506 |
| PPP1R12A | 97,8 | 95,9 | 100 | 99,8 | Genitourinary and/or/brain malformation syndrome, 618820 |
| PPP1R15B | 100 | 99,6 | 100 | 100 | Microcephaly, short stature, and impaired glucose metabolism 2, 616817 |
| PPP1R21 | 99,3 | 95,5 | 100 | 100 | Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 |
| PPP2CA | 100 | 100 | 100 | 100 | Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 |
| PPP2R1A | 91,6 | 91,6 | 93,6 | 93,6 | Mental retardation, autosomal dominant 36, 616362 |
| PPP2R5B | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PPP2R5C | 96,2 | 90,5 | 100 | 99,9 | No OMIM disease ID |

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|---|---|---|---|---|---|
| PPP2R5D | 100 | 99,8 | 100 | 100 | Mental retardation, autosomal dominant 35, 616355 |
| PPP3CA | 99,7 | 96,7 | 100 | 100 | Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265<br>Developmental and epileptic encephalopathy 91, 617711 |
| PPT1 | 90,3 | 89,9 | 82,5 | 82,5 | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PQBP1 | 100 | 99,3 | 100 | 100 | Renpenning syndrome, 309500 |
| PRDM15 | 99,6 | 96,9 | 99,7 | 98,8 | No OMIM disease ID |
| PRKACB | 98,5 | 96,1 | 100 | 99,9 | Cardioacrofacial dysplasia 2, 619143 |
| PRKAR1A | 97 | 89,1 | 100 | 100 | Pigmented nodular adrenocortical disease, primary, 1, 610489<br>Acrodysostosis 1, with or without hormone resistance, 101800<br>Carney complex, type 1, 160980<br>Myxoma, intracardiac, 255960<br>Adrenocortical tumor, somatic, |
| PRKAR1B | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PRMT7 | 100 | 99,9 | 100 | 100 | Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 |
| PRODH | 84 | 80,2 | 100 | 100 | Hyperprolinemia, type I, 239500 |
| PRPS1 | 86,4 | 86,3 | 100 | 99,7 | Arts syndrome, 301835<br>Phosphoribosylpyrophosphate synthetase superactivity, 300661<br>Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070<br>Deafness, X-linked 1, 304500<br>Gout, PRPS-related, 300661 |
| PRR12 | 98,4 | 96,7 | 100 | 100 | No OMIM disease ID |
| PRSS12 | 100 | 99,9 | 100 | 100 | Mental retardation, autosomal recessive 1, 249500 |
| PRUNE1 | 93,6 | 93,1 | 93,6 | 93,6 | Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 |
| PSAP | 100 | 99,6 | 100 | 100 | Combined SAP deficiency, 611721<br>Krabbe disease, atypical, 611722<br>Metachromatic leukodystrophy due to SAP-b deficiency, 249900<br>Gaucher disease, atypical, 610539 |
| PSAT1 | 92 | 75,1 | 100 | 100 | Neu-Laxova syndrome 2, 616038<br>?Phosphoserine aminotransferase deficiency, 610992 |
| PSMC5 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PSMD12 | 96,7 | 89,6 | 100 | 100 | Stankiewicz-Isidor syndrome, 617516 |
| PSPH | 100 | 100 | 100 | 100 | Phosphoserine phosphatase deficiency, 614023 |

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|---|---|---|---|---|---|
| PTCH1 | 99,3 | 96,6 | 100 | 99,9 | Basal cell carcinoma, somatic, 605462<br>Holoprosencephaly 7, 610828<br>Basal cell nevus syndrome, 109400 |
| PTCHD1 | 100 | 99,9 | 100 | 100 | No OMIM disease ID |
| PTDSS1 | 100 | 100 | 100 | 100 | Lenz-Majewski hyperostotic dwarfism, 151050 |
| PTEN | 99,5 | 97,2 | 100 | 100 | Lhermitte-Duclos syndrome, 158350<br>Cowden syndrome 1, 158350<br>Prostate cancer, somatic, 176807<br>Macrocephaly/autism syndrome, 605309 |
| PTF1A | 98,8 | 91,1 | 98,7 | 92,9 | Pancreatic and cerebellar agenesis, 609069<br>Pancreatic agenesis 2, 615935 |
| PTPN11 | 97,7 | 87,6 | 100 | 100 | Noonan syndrome 1, 163950<br>LEOPARD syndrome 1, 151100<br>Metachondromatosis, 156250<br>Leukemia, juvenile myelomonocytic, somatic, 607785 |
| PTPN23 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 |
| PTRH2 | 100 | 100 | 100 | 100 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 |
| PTRHD1 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PTS | 99,5 | 99 | 100 | 99,9 | Hyperphenylalaninemia, BH4-deficient, A, 261640 |
| PUF60 | 99,9 | 99,2 | 100 | 100 | Verheij syndrome, 615583 |
| PUM1 | 100 | 99,4 | 100 | 99,9 | Spinocerebellar ataxia 47, 617931 |
| PURA | 98,6 | 94,6 | 100 | 100 | Mental retardation, autosomal dominant 31, 616158 |
| PUS1 | 99,9 | 98 | 99,9 | 98,2 | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 |
| PUS3 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 |
| PUS7 | 99,8 | 99,7 | 100 | 100 | Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 |
| PYCR1 | 100 | 98,2 | 100 | 100 | Cutis laxa, autosomal recessive, type IIIB, 614438<br>Cutis laxa, autosomal recessive, type IIB, 612940 |
| PYCR2 | 100 | 99,3 | 100 | 100 | Leukodystrophy, hypomyelinating, 10, 616420 |
| QARS1 | 100 | 100 | 100 | 100 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 |
| QDPR | 100 | 98,9 | 100 | 100 | Hyperphenylalaninemia, BH4-deficient, C, 261630 |
| QRICH1 | 100 | 99 | 100 | 100 | Ververi-Brady syndrome, 617982 |

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|---|---|---|---|---|---|
| RAB11B | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 |
| RAB14 | 98,8 | 94,4 | 100 | 99,9 | No OMIM disease ID |
| RAB18 | 98,9 | 94,5 | 99,9 | 99,8 | Warburg micro syndrome 3, 614222 |
| RAB23 | 99,7 | 99,7 | 100 | 100 | Carpenter syndrome, 201000 |
| RAB27A | 99,5 | 99,5 | 100 | 99,9 | Griscelli syndrome, type 2, 607624 |
| RAB39B | 100 | 99,8 | 100 | 100 | Intellectual developmental disorder, X-linked 72, 300271<br>Waisman syndrome, 311510 |
| RAB3GAP1 | 99,2 | 98,7 | 99,4 | 99,3 | Martsolf syndrome 2, 619420<br>Warburg micro syndrome 1, 600118 |
| RAB3GAP2 | 99,1 | 96,3 | 100 | 99,9 | Martsolf syndrome 1, 212720<br>Warburg micro syndrome 2, 614225 |
| RAC1 | 99,6 | 96,3 | 100 | 100 | Mental retardation, autosomal dominant 48, 617751 |
| RAC3 | 98 | 94,3 | 99,4 | 97,3 | Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 |
| RAD21 | 99,2 | 95,9 | 100 | 100 | Cornelia de Lange syndrome 4, 614701<br>?Mungan syndrome, 611376 |
| RAF1 | 99,9 | 99,2 | 100 | 100 | Cardiomyopathy, dilated, 1NN, 615916<br>Noonan syndrome 5, 611553<br>LEOPARD syndrome 2, 611554 |
| RAI1 | 100 | 100 | 100 | 100 | Smith-Magenis syndrome, 182290 |
| RALA | 89,1 | 82,1 | 100 | 100 | Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 |
| RALGAPA1 | 73,6 | 61,2 | 100 | 99,9 | Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 |
| RARB | 100 | 99,8 | 100 | 100 | Microphthalmia, syndromic 12, 615524 |
| RARS1 | 94,1 | 91,8 | 94,4 | 94,2 | Leukodystrophy, hypomyelinating, 9, 616140 |
| RARS2 | 99,7 | 98,6 | 100 | 100 | Pontocerebellar hypoplasia, type 6, 611523 |
| RBBP8 | 99,7 | 99,4 | 100 | 99,9 | Seckel syndrome 2, 606744<br>Jawad syndrome, 251255<br>Pancreatic carcinoma, somatic, |
| RBFOX1 | 89,2 | 88,6 | 99,8 | 98,2 | No OMIM disease ID |
| RBM10 | 99,8 | 97,3 | 100 | 100 | TARP syndrome, 311900 |
| RBM28 | 100 | 100 | 100 | 100 | ?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 |
| RBPJ | 97,4 | 89 | 100 | 100 | Adams-Oliver syndrome 3, 614814 |

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|---|---|---|---|---|---|
| RCBTB1 | 99,7 | 98,6 | 100 | 100 | Retinal dystrophy with or without extraocular anomalies, 617175 |
| RECQL4 | 99,9 | 98,6 | 100 | 100 | Baller-Gerold syndrome, 218600<br>Rothmund-Thomson syndrome, type 2, 268400<br>RAPADILINO syndrome, 266280 |
| RELN | 100 | 99,6 | 100 | 100 | Lissencephaly 2 (Norman-Roberts type), 257320 |
| RERE | 93,3 | 85,2 | 99,9 | 99,9 | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 |
| REV3L | 97,4 | 97 | 97,6 | 97,6 | No OMIM disease ID |
| RFT1 | 99,7 | 98,4 | 100 | 100 | Congenital disorder of glycosylation, type In, 612015 |
| RFX3 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| RHEB | 86,5 | 70,4 | 100 | 100 | No OMIM disease ID |
| RHOBTB2 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 64, 618004 |
| RIC1 | 99,9 | 99,8 | 100 | 100 | CATIFA syndrome, 618761 |
| RIMS2 | 96,6 | 94,5 | 97,8 | 97,7 | Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 |
| RIT1 | 100 | 100 | 100 | 100 | Noonan syndrome 8, 615355 |
| RLIM | 99,8 | 98 | 100 | 100 | Tonne-Kalscheuer syndrome, 300978 |
| RMND1 | 99,7 | 97,2 | 100 | 99,9 | Combined oxidative phosphorylation deficiency 11, 614922 |
| RMRP | 0 | 0 | 0 | 0 | Anauxetic dysplasia 1, 607095<br>Metaphyseal dysplasia without hypotrichosis, 250460<br>Cartilage-hair hypoplasia, 250250 |
| RNASEH2A | 100 | 99,7 | 100 | 100 | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 81 | 78,2 | 91 | 90,9 | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 100 | 100 | 100 | 100 | Aicardi-Goutieres syndrome 3, 610329 |
| RNASET2 | 95,7 | 91 | 100 | 100 | Leukoencephalopathy, cystic, without megalencephaly, 612951 |
| RNF113A | 100 | 100 | 100 | 100 | Trichothiodystrophy 5, nonphotosensitive, 300953 |
| RNF125 | 100 | 98,3 | 100 | 100 | Tenorio syndrome, 616260 |
| RNF13 | 94,8 | 83,2 | 100 | 99,7 | Developmental and epileptic encephalopathy 73, 618379 |
| RNU4ATAC | 0 | 0 | 0 | 0 | Roifman syndrome, 616651<br>Lowry-Wood syndrome, 226960<br>Microcephalic osteodysplastic primordial dwarfism, type I, 210710 |
| ROGDI | 98,6 | 95,2 | 99,9 | 98,1 | Kohlschutter-Tonz syndrome, 226750 |

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|---|---|---|---|---|---|
| ROR2 | 100 | 99,4 | 97 | 97 | Brachydactyly, type B1, 113000<br>Robinow syndrome, autosomal recessive, 268310 |
| RORA | 96,2 | 89,4 | 100 | 100 | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 |
| RPGRIP1L | 96,5 | 95,3 | 100 | 99,4 | Joubert syndrome 7, 611560<br>Meckel syndrome 5, 611561<br>?COACH syndrome 3, 619113 |
| RPIA | 99,1 | 96,1 | 100 | 100 | Ribose 5-phosphate isomerase deficiency, 608611 |
| RPL10 | 96,7 | 87,5 | 100 | 100 | Intellectual developmental disorder, X-linked, syndromic, 35, 300998 |
| RPS19 | 100 | 99,9 | 100 | 100 | Diamond-Blackfan anemia 1, 105650 |
| RPS6KA3 | 98,4 | 91,4 | 99,9 | 98,3 | Intellectual developmental disorder, X-linked 19, 300844<br>Coffin-Lowry syndrome, 303600 |
| RRM2B | 100 | 99,8 | 100 | 99,9 | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075<br>Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075<br>Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 |
| RSPRY1 | 99,9 | 99,9 | 100 | 100 | Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 |
| RSRC1 | 99 | 94,6 | 100 | 100 | Intellectual developmental disorder, autosomal recessive 70, 618402 |
| RTEL1 | 99,7 | 97,2 | 100 | 100 | Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373<br>Dyskeratosis congenita, autosomal dominant 4, 615190<br>Dyskeratosis congenita, autosomal recessive 5, 615190 |
| RTN4IP1 | 99,6 | 97,3 | 100 | 100 | Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 |
| RTTN | 98,6 | 97,6 | 100 | 100 | Microcephaly, short stature, and polymicrogyria with seizures, 614833 |
| RUBCN | 99,7 | 97,9 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 15, 615705 |
| RUSC2 | 100 | 100 | 100 | 100 | Mental retardation, autosomal recessive 61, 617773 |
| RXYLT1 | 99,2 | 95,9 | 100 | 99,9 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 |
| SALL1 | 99,7 | 97,5 | 100 | 100 | Townes-Brocks syndrome 1, 107480<br>Townes-Brocks branchiootorenal-like syndrome, 107480 |
| SAMD9 | 99,9 | 99,8 | 100 | 100 | Tumoral calcinosis, familial, normophosphatemic, 610455<br>Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041<br>MIRAGE syndrome, 617053 |
| SAMHD1 | 98,5 | 97,9 | 100 | 100 | ?Chilblain lupus 2, 614415<br>Aicardi-Goutieres syndrome 5, 612952 |
| SARS1 | 100 | 99,3 | 100 | 100 | ?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 |
| SATB1 | 96,4 | 96 | 100 | 99,7 | Kohlschutter-Tonz syndrome-like, 619229<br>Developmental delay with dysmorphic facies and dental anomalies, 619228 |

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|---|---|---|---|---|---|
| SATB2 | 99,5 | 96,5 | 100 | 100 | Glass syndrome, 612313 |
| SBDS | 100 | 99,9 | 100 | 100 | Shwachman-Diamond syndrome, 260400 |
| SC5D | 99,9 | 99,1 | 100 | 100 | Lathosterolosis, 607330 |
| SCAF4 | 99,3 | 97,1 | 100 | 100 | No OMIM disease ID |
| SCAMP5 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| SCAPER | 99,5 | 97,1 | 100 | 99,9 | Intellectual developmental disorder and retinitis pigmentosa, 618195 |
| SCN1A | 99,7 | 99,1 | 100 | 100 | Developmental and epileptic encephalopathy 6B, non-Dravet, 619317<br>Migraine, familial hemiplegic, 3, 609634<br>Dravet syndrome, 607208<br>Febrile seizures, familial, 3A, 604403<br>Generalized epilepsy with febrile seizures plus, type 2, 604403 |
| SCN1B | 98,2 | 96,3 | 99,7 | 98,9 | Generalized epilepsy with febrile seizures plus, type 1, 604233<br>Developmental and epileptic encephalopathy 52, 617350<br>Cardiac conduction defect, nonspecific, 612838<br>Atrial fibrillation, familial, 13, 615377<br>Brugada syndrome 5, 612838 |
| SCN2A | 99,4 | 97,4 | 100 | 99,9 | Seizures, benign familial infantile, 3, 607745<br>Developmental and epileptic encephalopathy 11, 613721<br>Episodic ataxia, type 9, 618924 |
| SCN3A | 99,8 | 99,1 | 100 | 100 | Epilepsy, familial focal, with variable foci 4, 617935<br>Developmental and epileptic encephalopathy 62, 617938 |
| SCN8A | 100 | 99,5 | 100 | 100 | ?Myoclonus, familial, 2, 618364<br>Seizures, benign familial infantile, 5, 617080<br>Cognitive impairment with or without cerebellar ataxia, 614306<br>Developmental and epileptic encephalopathy 13, 614558 |
| SCO1 | 97,6 | 94,4 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 4, 619048 |
| SCO2 | 100 | 100 | 100 | 100 | Myopia 6, 608908<br>Mitochondrial complex IV deficiency, nuclear type 2, 604377 |
| SCUBE3 | 100 | 99,8 | 100 | 100 | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184 |
| SCYL1 | 100 | 99,9 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 21, 616719 |
| SDCCAG8 | 99,8 | 99,8 | 100 | 100 | Senior-Loken syndrome 7, 613615<br>Bardet-Biedl syndrome 16, 615993 |
| SDHA | 84,5 | 77,9 | 100 | 100 | Cardiomyopathy, dilated, 1GG, 613642<br>Mitochondrial complex II deficiency, nuclear type 1, 252011 |

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| | | | | | Neurodegeneration with ataxia and late-onset optic atrophy, 619259<br>Paragangliomas 5, 614165 |
| SEC31A | 99 | 96,2 | 100 | 100 | ?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651 |
| SEMA3E | 99,1 | 98,9 | 100 | 100 | ?CHARGE syndrome, 214800 |
| SEPSECS | 99,9 | 99,6 | 100 | 100 | Pontocerebellar hypoplasia type 2D, 613811 |
| SERAC1 | 99,6 | 99,5 | 100 | 99,9 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |
| SET | 96,7 | 87,4 | 99,3 | 97,3 | Mental retardation, autosomal dominant 58, 618106 |
| SETBP1 | 99,5 | 98,3 | 100 | 100 | Schinzel-Giedion midface retraction syndrome, 269150<br>Mental retardation, autosomal dominant 29, 616078 |
| SETD1A | 100 | 99,7 | 100 | 100 | Epilepsy, early-onset, with or without developmental delay, 618832<br>Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 |
| SETD1B | 98,3 | 97,6 | 100 | 100 | Intellectual developmental disorder with seizures and language delay, 619000 |
| SETD2 | 99,9 | 99,6 | 100 | 100 | Luscan-Lumish syndrome, 616831 |
| SETD5 | 100 | 99,7 | 98 | 98 | Mental retardation, autosomal dominant 23, 615761 |
| SFXN4 | 99,6 | 97,4 | 100 | 100 | Combined oxidative phosphorylation deficiency 18, 615578 |
| SGPL1 | 100 | 100 | 100 | 100 | Nephrotic syndrome, type 14, 617575 |
| SGSH | 94,8 | 94,1 | 100 | 100 | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 |
| SHANK2 | 97,7 | 97,5 | 98,9 | 98,9 | No OMIM disease ID |
| SHANK3 | 92,4 | 84,9 | 96 | 91,6 | Phelan-McDermid syndrome, 606232 |
| SHH | 100 | 100 | 100 | 100 | Microphthalmia with coloboma 5, 611638<br>Schizencephaly, 269160<br>Single median maxillary central incisor, 147250<br>Holoprosencephaly 3, 142945 |
| SHMT2 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 |
| SHOC2 | 99,8 | 99,6 | 100 | 99,9 | Noonan syndrome-like with loose anagen hair 1, 607721 |
| SHROOM4 | 99,8 | 98,1 | 100 | 100 | Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434 |
| SIAH1 | 100 | 99,9 | 100 | 100 | Buratti-Harel syndrome, 619314 |
| SIK1 | 98,2 | 93,9 | 100 | 100 | Developmental and epileptic encephalopathy 30, 616341 |
| SIL1 | 98,7 | 96 | 100 | 100 | Marinesco-Sjogren syndrome, 248800 |
| SIN3A | 99,8 | 98,2 | 100 | 100 | Witteveen-Kolk syndrome, 613406 |
| SIN3B | 96,6 | 96 | 100 | 100 | No OMIM disease ID |

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|---|---|---|---|---|---|
| SIX3 | 99,3 | 96,9 | 100 | 99,8 | Schizencephaly, 269160<br>Holoprosencephaly 2, 157170 |
| SKI | 99,7 | 97,1 | 100 | 99,7 | Shprintzen-Goldberg syndrome, 182212 |
| SLC12A2 | 94,4 | 92,4 | 100 | 100 | Kilquist syndrome, 619080<br>Delpire-McNeill syndrome, 619083<br>Deafness, autosomal dominant 78, 619081 |
| SLC12A5 | 83,9 | 83,8 | 97,4 | 97,4 | Developmental and epileptic encephalopathy 34, 616645 |
| SLC12A6 | 100 | 100 | 100 | 100 | Agenesis of the corpus callosum with peripheral neuropathy, 218000 |
| SLC13A5 | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 |
| SLC16A2 | 97,6 | 88,4 | 100 | 100 | Allan-Herndon-Dudley syndrome, 300523 |
| SLC17A5 | 99,6 | 96,2 | 100 | 100 | Salla disease, 604369<br>Sialic acid storage disorder, infantile, 269920 |
| SLC19A3 | 97,8 | 97 | 98,7 | 98,7 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 |
| SLC1A1 | 100 | 99,5 | 100 | 100 | Dicarboxylic aminoaciduria, 222730 |
| SLC1A2 | 96,1 | 94,8 | 100 | 100 | Developmental and epileptic encephalopathy 41, 617105 |
| SLC1A4 | 99,6 | 97 | 100 | 100 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 |
| SLC25A1 | 96,9 | 89,8 | 99,7 | 98,2 | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182<br>Myasthenic syndrome, congenital, 23, presynaptic, 618197 |
| SLC25A12 | 100 | 99,2 | 100 | 100 | Developmental and epileptic encephalopathy 39, 612949 |
| SLC25A15 | 99,3 | 96,6 | 100 | 100 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A22 | 99,2 | 96,5 | 100 | 100 | Developmental and epileptic encephalopathy 3, 609304 |
| SLC25A24 | 99,3 | 98,8 | 99,7 | 99,7 | Fontaine progeroid syndrome, 612289 |
| SLC25A42 | 97,1 | 94,3 | 100 | 100 | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 |
| SLC2A1 | 92,8 | 92,7 | 100 | 100 | Dystonia 9, 601042<br>GLUT1 deficiency syndrome 1, infantile onset, severe, 606777<br>Stomatin-deficient cryohydrocytosis with neurologic defects, 608885<br>GLUT1 deficiency syndrome 2, childhood onset, 612126 |
| SLC33A1 | 99,8 | 98,5 | 100 | 99,8 | Spastic paraplegia 42, autosomal dominant, 612539<br>Congenital cataracts, hearing loss, and neurodegeneration, 614482 |
| SLC35A1 | 99,7 | 99,3 | 100 | 100 | Congenital disorder of glycosylation, type IIf, 603585 |
| SLC35A2 | 99,6 | 97,7 | 100 | 100 | Congenital disorder of glycosylation, type IIm, 300896 |

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| SLC35A3 | 80,4 | 78,8 | 81 | 80,9 | ?Arthrogryposis, mental retardation, and seizures, 615553 |
| SLC35C1 | 100 | 99,4 | 100 | 100 | Congenital disorder of glycosylation, type IIc, 266265 |
| SLC39A14 | 100 | 99 | 93,5 | 93,5 | ?Hyperostosis cranalis interna, 144755<br>Hypermanganesemia with dystonia 2, 617013 |
| SLC39A8 | 100 | 99,7 | 100 | 100 | Congenital disorder of glycosylation, type IIn, 616721 |
| SLC46A1 | 100 | 98,5 | 100 | 100 | Folate malabsorption, hereditary, 229050 |
| SLC4A4 | 99,9 | 99,4 | 100 | 100 | Renal tubular acidosis, proximal, with ocular abnormalities, 604278 |
| SLC5A6 | 100 | 100 | 100 | 100 | Neurodegeneration, infantile-onset, biotin-responsive, 618973 |
| SLC6A1 | 96,7 | 96,6 | 100 | 100 | Myoclonic-atonic epilepsy, 616421 |
| SLC6A17 | 100 | 100 | 100 | 100 | Mental retardation, autosomal recessive 48, 616269 |
| SLC6A19 | 100 | 100 | 100 | 100 | Iminoglycinuria, digenic, 242600<br>Hartnup disorder, 234500<br>Hyperglycinuria, 138500 |
| SLC6A3 | 100 | 99,9 | 100 | 100 | Parkinsonism-dystonia, infantile, 1, 613135 |
| SLC6A8 | 94,8 | 83 | 99,9 | 99,5 | Cerebral creatine deficiency syndrome 1, 300352 |
| SLC6A9 | 100 | 99,6 | 100 | 100 | Glycine encephalopathy with normal serum glycine, 617301 |
| SLC7A7 | 100 | 99,9 | 100 | 100 | Lysinuric protein intolerance, 222700 |
| SLC9A6 | 94,7 | 90,2 | 99,7 | 97 | Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 |
| SLC9A7 | 97,4 | 89,8 | 100 | 99,6 | Intellectual developmental disorder, X-linked 108, 301024 |
| SMAD4 | 99,9 | 99,9 | 100 | 100 | Pancreatic cancer, somatic, 260350<br>Myhre syndrome, 139210<br>Polyposis, juvenile intestinal, 174900<br>Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 |
| SMARCA1 | 99,1 | 96,3 | 99,9 | 98,8 | No OMIM disease ID |
| SMARCA2 | 96,7 | 96,3 | 97,8 | 96,9 | Nicolaides-Baraitser syndrome, 601358<br>Blepharophimosis-impaired intellectual development syndrome, 619293 |
| SMARCA4 | 99,9 | 99,2 | 100 | 100 | Coffin-Siris syndrome 4, 614609 |
| SMARCA5 | 99,3 | 96,9 | 100 | 99,9 | No OMIM disease ID |
| SMARCB1 | 100 | 99,9 | 100 | 100 | Rhabdoid tumors, somatic, 609322<br>Coffin-Siris syndrome 3, 614608 |
| SMARCC2 | 98,8 | 95,7 | 100 | 100 | Coffin-Siris syndrome 8, 618362 |
| SMARCD1 | 94,9 | 89,8 | 100 | 99,9 | Coffin-Siris syndrome 11, 618779 |

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| SMARCE1 | 93,7 | 85,9 | 100 | 100 | Coffin-Siris syndrome 5, 616938 |
| SMC1A | 99,6 | 97,1 | 100 | 99,9 | Cornelia de Lange syndrome 2, 300590<br>Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 |
| SMC3 | 94,5 | 89 | 100 | 99,9 | Cornelia de Lange syndrome 3, 610759 |
| SMG8 | 100 | 100 | 100 | 100 | Alzahrani-Kuwahara syndrome, 619268 |
| SMG9 | 100 | 100 | 100 | 100 | Heart and brain malformation syndrome, 616920 |
| SMOC1 | 99,8 | 98,2 | 100 | 100 | Microphthalmia with limb anomalies, 206920 |
| SMPD1 | 100 | 99,9 | 100 | 100 | Niemann-Pick disease, type B, 607616<br>Niemann-Pick disease, type A, 257200 |
| SMPD4 | 99,6 | 95 | 100 | 100 | Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 |
| SMS | 87,9 | 72,1 | 100 | 99,5 | Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 |
| SNAP25 | 100 | 99,8 | 100 | 100 | ?Myasthenic syndrome, congenital, 18, 616330 |
| SNAP29 | 100 | 100 | 100 | 100 | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 |
| SNIP1 | 99,2 | 97,3 | 100 | 100 | Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 |
| SNORD118 | 0 | 0 | 0 | 0 | Leukoencephalopathy, brain calcifications, and cysts, 614561 |
| SNRPB | 100 | 98,6 | 100 | 100 | Cerebrocostomandibular syndrome, 117650 |
| SNRPN | 99,8 | 96,5 | 100 | 100 | Prader-Willi syndrome, 176270 |
| SNX14 | 98,9 | 93,6 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 20, 616354 |
| SNX27 | 100 | 99,1 | 100 | 100 | No OMIM disease ID |
| SOBP | 98,5 | 95,9 | 97,4 | 95,5 | Mental retardation, anterior maxillary protrusion, and strabismus, 613671 |
| SON | 97,6 | 92,6 | 100 | 100 | ZTTK syndrome, 617140 |
| SOS1 | 99,6 | 97,9 | 100 | 99,9 | Noonan syndrome 4, 610733<br>?Fibromatosis, gingival, 1, 135300 |
| SOS2 | 99,6 | 98,7 | 100 | 99,9 | Noonan syndrome 9, 616559 |
| SOX10 | 99,9 | 97,2 | 100 | 100 | Waardenburg syndrome, type 4C, 613266<br>PCWH syndrome, 609136<br>Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 |
| SOX11 | 100 | 100 | 100 | 100 | Coffin-Siris syndrome 9, 615866 |
| SOX2 | 100 | 99,8 | 100 | 100 | Optic nerve hypoplasia and abnormalities of the central nervous system, 206900<br>Microphthalmia, syndromic 3, 206900 |

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| SOX3 | 94,9 | 81,2 | 100 | 99,6 | Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123<br>Panhypopituitarism, X-linked, 312000 |
| SOX4 | 97,7 | 90,6 | 99,8 | 97,9 | Coffin-Siris syndrome 10, 618506 |
| SOX5 | 99,8 | 97,6 | 100 | 100 | Lamb-Shaffer syndrome, 616803 |
| SOX6 | 99,9 | 98,9 | 100 | 99,9 | Tolchin-Le Caignec syndrome, 618971 |
| SPART | 99,7 | 96,4 | 100 | 100 | Troyer syndrome, 275900 |
| SPAST | 99,4 | 98,1 | 100 | 100 | Spastic paraplegia 4, autosomal dominant, 182601 |
| SPATA5 | 99,8 | 99,5 | 100 | 100 | Epilepsy, hearing loss, and mental retardation syndrome, 616577 |
| SPECC1L | 96 | 95 | 97,1 | 96,1 | Opitz GBBB syndrome, type II, 145410<br>Teebi hypertelorism syndrome, 145420<br>?Facial clefting, oblique, 1, 600251 |
| SPEN | 100 | 99,8 | 100 | 100 | Radio-Tartaglia syndrome, 619312 |
| SPG11 | 99,8 | 99 | 100 | 100 | Amyotrophic lateral sclerosis 5, juvenile, 602099<br>Charcot-Marie-Tooth disease, axonal, type 2X, 616668<br>Spastic paraplegia 11, autosomal recessive, 604360 |
| SPOCK1 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| SPOP | 100 | 100 | 100 | 100 | Nabais Sa-de Vries syndrome, type 1, 618828<br>Nabais Sa-de Vries syndrome, type 2, 618829 |
| SPR | 100 | 99,4 | 100 | 100 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 |
| SPRED1 | 99,8 | 98,2 | 100 | 100 | Legius syndrome, 611431 |
| SPTAN1 | 99,1 | 97,9 | 100 | 100 | Developmental and epileptic encephalopathy 5, 613477 |
| SPTBN1 | 99,9 | 99,4 | 100 | 100 | Developmental delay, impaired speech, and behavioral abnormalities, 619475 |
| SPTBN2 | 100 | 99,4 | 100 | 99,9 | Spinocerebellar ataxia 5, 600224<br>Spinocerebellar ataxia, autosomal recessive 14, 615386 |
| SPTBN4 | 98,1 | 92,1 | 100 | 100 | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 |
| SRCAP | 99,7 | 98,9 | 100 | 100 | Floating-Harbor syndrome, 136140 |
| SRD5A3 | 100 | 99,1 | 100 | 100 | Kahrizi syndrome, 612713<br>Congenital disorder of glycosylation, type Iq, 612379 |
| SRPX2 | 99,3 | 93,6 | 100 | 100 | ?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643 |
| SRRM2 | 100 | 99,8 | 100 | 100 | No OMIM disease ID |
| SSR4 | 100 | 99,5 | 100 | 100 | Congenital disorder of glycosylation, type Iy, 300934 |
| ST3GAL3 | 68,8 | 68,2 | 95,3 | 95,2 | Developmental and epileptic encephalopathy 15, 615006<br>Intellectual developmental disorder, autosomal recessive 12, 611090 |

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| ST3GAL5 | 85,9 | 84 | 98,7 | 98,6 | Salt and pepper developmental regression syndrome, 609056 |
| STAG1 | 99,4 | 96,2 | 100 | 100 | Mental retardation, autosomal dominant 47, 617635 |
| STAG2 | 97 | 86,9 | 100 | 99,3 | Holoprosencephaly 13, X-linked, 301043<br>Mullegama-Klein-Martinez syndrome, 301022 |
| STAMBP | 99,4 | 96,4 | 100 | 100 | Microcephaly-capillary malformation syndrome, 614261 |
| CXorf56 | 99,4 | 92,9 | 100 | 99,8 | ?Intellectual developmental disorder, X-linked 107, 301013 |
| STIL | 99,9 | 99,7 | 100 | 100 | Microcephaly 7, primary, autosomal recessive, 612703 |
| STRA6 | 100 | 99,9 | 100 | 100 | Microphthalmia, syndromic 9, 601186<br>Microphthalmia, isolated, with coloboma 8, 601186 |
| STRADA | 100 | 99 | 100 | 100 | Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 |
| STT3A | 100 | 100 | 100 | 100 | Congenital disorder of glycosylation, type Iw, 615596 |
| STT3B | 99,7 | 99,4 | 100 | 100 | ?Congenital disorder of glycosylation, type Ix, 615597 |
| STX1B | 100 | 100 | 100 | 100 | Generalized epilepsy with febrile seizures plus, type 9, 616172 |
| STXBP1 | 96,8 | 96,2 | 100 | 100 | Developmental and epileptic encephalopathy 4, 612164 |
| SUCLA2 | 88,8 | 79,4 | 99,9 | 99,8 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| SUCLG1 | 100 | 99,7 | 100 | 99,8 | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 |
| SUMF1 | 98,3 | 92,5 | 100 | 100 | Multiple sulfatase deficiency, 272200 |
| SUOX | 100 | 100 | 100 | 100 | Sulfite oxidase deficiency, 272300 |
| SUPT16H | 97 | 89,3 | 100 | 100 | Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 |
| SURF1 | 89,5 | 88,1 | 100 | 100 | Charcot-Marie-Tooth disease, type 4K, 616684<br>Mitochondrial complex IV deficiency, nuclear type 1, 220110 |
| SUZ12 | 90,7 | 86,2 | 100 | 99,9 | Imagawa-Matsumoto syndrome, 618786 |
| SVBP | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 |
| SYN1 | 82 | 71,6 | 100 | 99,9 | Intellectual developmental disorder, X-linked 50, 300115<br>Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 |
| SYNCRIP | 97 | 82 | 100 | 100 | No OMIM disease ID |
| SYNGAP1 | 98,8 | 97,4 | 100 | 100 | Mental retardation, autosomal dominant 5, 612621 |
| SYNJ1 | 99,7 | 98,1 | 100 | 100 | Parkinson disease 20, early-onset, 615530<br>Developmental and epileptic encephalopathy 53, 617389 |
| SYP | 99,9 | 96,2 | 100 | 100 | Intellectual developmental disorder, X-linked 96, 300802 |

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| SYT1 | 99,5 | 97,5 | 100 | 100 | Baker-Gordon syndrome, 618218 |
| SZT2 | 99,6 | 99,3 | 100 | 99,9 | Developmental and epileptic encephalopathy 18, 615476 |
| TACO1 | 98,9 | 93,7 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 8, 619052 |
| TAF1 | 99,2 | 95,7 | 100 | 100 | Intellectual developmental disorder, X-linked syndromic 33, 300966<br>Dystonia-Parkinsonism, X-linked, 314250 |
| TAF13 | 99,6 | 99,1 | 100 | 99,9 | Mental retardation, autosomal recessive 60, 617432 |
| TAF1C | 100 | 100 | 100 | 100 | No OMIM disease ID |
| TAF2 | 99,5 | 98,6 | 100 | 99,9 | Mental retardation, autosomal recessive 40, 615599 |
| TAF6 | 99,6 | 98,1 | 100 | 100 | Alazami-Yuan syndrome, 617126 |
| TANC2 | 99,8 | 98,9 | 100 | 100 | Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 |
| TANGO2 | 100 | 99,3 | 100 | 100 | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| TAOK1 | 99,6 | 97,5 | 100 | 100 | No OMIM disease ID |
| TASP1 | 99,8 | 98,7 | 100 | 100 | Suleiman-El-Hattab syndrome, 618950 |
| TAT | 100 | 100 | 100 | 100 | Tyrosinemia, type II, 276600 |
| TBC1D20 | 94,3 | 93,9 | 100 | 99,7 | Warburg micro syndrome 4, 615663 |
| TBC1D23 | 98,7 | 94,5 | 100 | 99,7 | Pontocerebellar hypoplasia, type 11, 617695 |
| TBC1D24 | 100 | 100 | 100 | 100 | Deafness, autosomal recessive 86, 614617<br>Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105<br>Myoclonic epilepsy, infantile, familial, 605021<br>Deafness, autosomal dominant 65, 616044<br>Developmental and epileptic encephalopathy 16, 615338<br>DOORS syndrome, 220500 |
| TBC1D2B | 99 | 97,1 | 98,5 | 97,9 | Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 |
| TBC1D7 | 99,7 | 99,3 | 100 | 100 | Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 |
| TBCD | 95,5 | 93,3 | 100 | 100 | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 |
| TBCE | 99,7 | 96,6 | 100 | 100 | Kenny-Caffey syndrome, type 1, 244460<br>Hypoparathyroidism-retardation-dysmorphism syndrome, 241410<br>Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 |
| TBCK | 99,4 | 95,8 | 100 | 99,9 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 |
| TBL1XR1 | 93,4 | 80,7 | 100 | 100 | Pierpont syndrome, 602342<br>Mental retardation, autosomal dominant 41, 616944 |

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| TBP | 99,9 | 99,2 | 100 | 99,9 | Spinocerebellar ataxia 17, 607136 |
| TBR1 | 100 | 99,6 | 100 | 100 | Intellectual developmental disorder with autism and speech delay, 606053 |
| TBX1 | 87,4 | 77,6 | 93,7 | 90,2 | Tetralogy of Fallot, 187500<br>DiGeorge syndrome, 188400<br>Conotruncal anomaly face syndrome, 217095<br>Velocardiofacial syndrome, 192430 |
| TCF20 | 100 | 100 | 100 | 100 | Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 |
| TCF4 | 100 | 99,9 | 100 | 100 | Pitt-Hopkins syndrome, 610954<br>Corneal dystrophy, Fuchs endothelial, 3, 613267 |
| TCF7L2 | 99,3 | 97 | 100 | 100 | No OMIM disease ID |
| TCN2 | 100 | 100 | 100 | 100 | Transcobalamin II deficiency, 275350 |
| TCTN2 | 99,9 | 99,1 | 100 | 100 | Joubert syndrome 24, 616654<br>?Meckel syndrome 8, 613885 |
| TCTN3 | 100 | 100 | 100 | 100 | Joubert syndrome 18, 614815<br>Orofaciodigital syndrome IV, 258860 |
| TDP2 | 99,6 | 99,5 | 100 | 99,9 | Spinocerebellar ataxia, autosomal recessive 23, 616949 |
| TECPR2 | 100 | 100 | 100 | 100 | Spastic paraplegia 49, autosomal recessive, 615031 |
| TECR | 100 | 98,5 | 100 | 100 | Mental retardation, autosomal recessive 14, 614020 |
| TELO2 | 99,9 | 98 | 100 | 100 | You-Hoover-Fong syndrome, 616954 |
| TENM3 | 100 | 99,7 | 100 | 100 | Microphthalmia, syndromic 15, 615145<br>?Microphthalmia, isolated, with coloboma 9, 615145 |
| TET3 | 94,4 | 94,4 | 100 | 100 | Beck-Fahrner syndrome, 618798 |
| TFAP2A | 98,1 | 92,1 | 100 | 100 | Branchiooculofacial syndrome, 113620 |
| TFE3 | 98,1 | 91 | 100 | 100 | Renal cell carcinoma, papillary, 1, 300854 |
| TGDS | 99,4 | 95,9 | 100 | 99,9 | Catel-Manzke syndrome, 616145 |
| TGFBR1 | 93,6 | 93,6 | 98,8 | 97,6 | Loeys-Dietz syndrome 1, 609192 |
| TGIF1 | 100 | 100 | 100 | 100 | Holoprosencephaly 4, 142946 |
| TH | 99,8 | 98 | 100 | 100 | Segawa syndrome, recessive, 605407 |
| THOC2 | 98,1 | 91,2 | 100 | 99,5 | Intellectual developmental disorder, X-linked 12, 300957 |
| THOC6 | 100 | 100 | 100 | 100 | Beaulieu-Boycott-Innes syndrome, 613680 |

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|---|---|---|---|---|---|
| THRB | 100 | 99,6 | 100 | 100 | Thyroid hormone resistance, autosomal recessive, 274300<br>Thyroid hormone resistance, 188570<br>Thyroid hormone resistance, selective pituitary, 145650 |
| TIMM50 | 98,4 | 95 | 100 | 100 | 3-methylglutaconic aciduria, type IX, 617698 |
| TIMM8A | 96,2 | 83,1 | 100 | 100 | Mohr-Tranebjaerg syndrome, 304700 |
| TINF2 | 100 | 100 | 100 | 100 | Dyskeratosis congenita, autosomal dominant 3, 613990<br>Revesz syndrome, 268130 |
| TKFC | 100 | 99,8 | 100 | 100 | Triokinase and FMN cyclase deficiency syndrome, 618805 |
| TKT | 98,6 | 96,8 | 98,7 | 98,7 | Short stature, developmental delay, and congenital heart defects, 617044 |
| TLK2 | 98,5 | 93,2 | 100 | 100 | Mental retardation, autosomal dominant 57, 618050 |
| TMCO1 | 87,8 | 87 | 88 | 87,9 | Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 |
| TMEM106B | 99,3 | 98,3 | 100 | 99,9 | Leukodystrophy, hypomyelinating, 16, 617964 |
| TMEM165 | 100 | 100 | 100 | 100 | Congenital disorder of glycosylation, type IIk, 614727 |
| TMEM216 | 98,5 | 92,8 | 100 | 100 | Joubert syndrome 2, 608091<br>Meckel syndrome 2, 603194 |
| TMEM222 | 100 | 99,5 | 100 | 100 | Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 |
| TMEM231 | 100 | 99,3 | 100 | 100 | Joubert syndrome 20, 614970<br>Meckel syndrome 11, 615397 |
| TMEM237 | 99,8 | 99,3 | 100 | 100 | Joubert syndrome 14, 614424 |
| TMEM240 | 100 | 100 | 100 | 100 | Spinocerebellar ataxia 21, 607454 |
| TMEM63A | 100 | 99,2 | 100 | 100 | Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 |
| TMEM67 | 98,6 | 93,5 | 100 | 99,6 | Nephronophthisis 11, 613550<br>Joubert syndrome 6, 610688<br>Meckel syndrome 3, 607361<br>?RHYNS syndrome, 602152<br>COACH syndrome 1, 216360 |
| TMEM70 | 98,4 | 94,6 | 100 | 100 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 |
| TMEM94 | 100 | 100 | 100 | 100 | Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 |
| TMLHE | 98,6 | 94,1 | 100 | 99,7 | No OMIM disease ID |
| TMTC3 | 98,7 | 95,8 | 100 | 100 | Lissencephaly 8, 617255 |
| TMX2 | 100 | 99,2 | 100 | 100 | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 |
| TNIK | 99,8 | 98,5 | 100 | 100 | Mental retardation, autosomal recessive 54, 617028 |

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|---|---|---|---|---|---|
| TNR | 100 | 99,6 | 100 | 100 | No OMIM disease ID |
| TNRC6B | 99,9 | 99,2 | 100 | 100 | Global developmental delay with speech and behavioral abnormalities, 619243 |
| TOE1 | 100 | 100 | 100 | 100 | Pontocerebellar hypoplasia, type 7, 614969 |
| TOGARAM1 | 99,6 | 97,5 | 100 | 99,9 | Joubert syndrome 37, 619185 |
| TOMM70 | 99,9 | 99,3 | 100 | 100 | No OMIM disease ID |
| TOR1A | 91,3 | 91,3 | 91,7 | 91,3 | Arthrogryposis multiplex congenita 5, 618947<br>Dystonia-1, torsion, 128100 |
| TP53RK | 95,5 | 84,9 | 100 | 100 | Galloway-Mowat syndrome 4, 617730 |
| TPI1 | 99,8 | 98 | 100 | 100 | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 |
| TPO | 100 | 99,2 | 100 | 100 | Thyroid dyshormonogenesis 2A, 274500 |
| TPP1 | 100 | 100 | 100 | 100 | Ceroid lipofuscinosis, neuronal, 2, 204500<br>Spinocerebellar ataxia, autosomal recessive 7, 609270 |
| TPP2 | 99,2 | 95,1 | 100 | 100 | Immunodeficiency 78 with autoimmunity and developmental delay, 619220 |
| TPRKB | 80,2 | 75,2 | 81,9 | 81,7 | Galloway-Mowat syndrome 5, 617731 |
| TRAF7 | 100 | 100 | 100 | 100 | Cardiac, facial, and digital anomalies with developmental delay, 618164 |
| TRAIP | 100 | 100 | 100 | 99,9 | Seckel syndrome 9, 616777 |
| TRAK1 | 93,3 | 93,1 | 100 | 99,9 | Developmental and epileptic encephalopathy 68, 618201 |
| TRAPPC11 | 99,7 | 98,7 | 100 | 99,9 | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 |
| TRAPPC2L | 100 | 100 | 100 | 100 | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 |
| TRAPPC4 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 |
| TRAPPC6B | 99,2 | 96,4 | 100 | 100 | Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 |
| TRAPPC9 | 100 | 99,7 | 100 | 100 | Mental retardation, autosomal recessive 13, 613192 |
| TREX1 | 100 | 100 | 100 | 100 | Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315<br>Aicardi-Goutieres syndrome 1, dominant and recessive, 225750<br>Chilblain lupus, 610448 |
| TRIM32 | 100 | 99,9 | 100 | 100 | ?Bardet-Biedl syndrome 11, 615988<br>Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 |
| TRIM8 | 98,9 | 96,2 | 100 | 100 | Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 |
| TRIO | 99,3 | 97,5 | 99,2 | 98,2 | Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061<br>Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 |
| TRIP12 | 99,7 | 98,6 | 100 | 100 | Mental retardation, autosomal dominant 49, 617752 |

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|---|---|---|---|---|---|
| TRIT1 | 100 | 100 | 100 | 100 | Combined oxidative phosphorylation deficiency 35, 617873 |
| TRMT1 | 99,5 | 96,2 | 100 | 100 | Mental retardation, autosomal recessive 68, 618302 |
| TRMT10A | 99,7 | 99,5 | 100 | 100 | Microcephaly, short stature, and impaired glucose metabolism 1, 616033 |
| TRNT1 | 99,7 | 97,4 | 100 | 99,9 | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084<br>Retinitis pigmentosa and erythrocytic microcytosis, 616959 |
| TRPM3 | 99,9 | 99,4 | 100 | 100 | No OMIM disease ID |
| TRRAP | 99,9 | 99,1 | 100 | 100 | ?Deafness, autosomal dominant 75, 618778<br>Developmental delay with or without dysmorphic facies and autism, 618454 |
| TSC1 | 99,5 | 98,2 | 100 | 100 | Focal cortical dysplasia, type II, somatic, 607341<br>Tuberous sclerosis-1, 191100<br>Lymphangioleiomyomatosis, 606690 |
| TSC2 | 100 | 99,8 | 100 | 100 | Lymphangioleiomyomatosis, somatic, 606690<br>?Focal cortical dysplasia, type II, somatic, 607341<br>Tuberous sclerosis-2, 613254 |
| TSEN15 | 78,9 | 77 | 100 | 100 | Pontocerebellar hypoplasia, type 2F, 617026 |
| TSEN2 | 99,9 | 99,2 | 100 | 100 | Pontocerebellar hypoplasia type 2B, 612389 |
| TSEN54 | 96,7 | 94,8 | 99,9 | 99,2 | Pontocerebellar hypoplasia type 2A, 277470<br>Pontocerebellar hypoplasia type 4, 225753<br>?Pontocerebellar hypoplasia type 5, 610204 |
| TSFM | 100 | 99,3 | 94,9 | 94,9 | Combined oxidative phosphorylation deficiency 3, 610505 |
| TSHB | 100 | 100 | 100 | 100 | Hypothyroidism, congenital, nongoitrous 4, 275100 |
| TSPAN7 | 100 | 99,8 | 100 | 100 | Intellectual developmental disorder, X-linked 58, 300210 |
| TTC19 | 83,8 | 74,1 | 100 | 99,8 | Mitochondrial complex III deficiency, nuclear type 2, 615157 |
| TTC37 | 99,7 | 98,8 | 100 | 100 | Trichohepatoenteric syndrome 1, 222470 |
| TTC5 | 99,9 | 99 | 100 | 100 | Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 |
| TTC8 | 99,5 | 98 | 100 | 100 | Bardet-Biedl syndrome 8, 615985<br>?Retinitis pigmentosa 51, 613464 |
| TTI2 | 100 | 99,9 | 100 | 100 | Mental retardation, autosomal recessive 39, 615541 |
| TUBA1A | 99,5 | 93,2 | 100 | 100 | Lissencephaly 3, 611603 |
| TUBA8 | 99,9 | 99,2 | 100 | 100 | No OMIM disease ID |
| TUBB | 96,8 | 93,7 | 99,9 | 99,8 | Symmetric circumferential skin creases, congenital, 1, 156610<br>Cortical dysplasia, complex, with other brain malformations 6, 615771 |
| TUBB2A | 96,9 | 95,7 | 100 | 100 | Cortical dysplasia, complex, with other brain malformations 5, 615763 |

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|---|---|---|---|---|---|
| TUBB2B | 100 | 99,7 | 100 | 100 | Cortical dysplasia, complex, with other brain malformations 7, 610031 |
| TUBB3 | 98,5 | 96,8 | 100 | 100 | Fibrosis of extraocular muscles, congenital, 3A, 600638<br>Cortical dysplasia, complex, with other brain malformations 1, 614039 |
| TUBB4A | 96 | 95,6 | 99 | 96,9 | Dystonia 4, torsion, autosomal dominant, 128101<br>Leukodystrophy, hypomyelinating, 6, 612438 |
| TUBG1 | 100 | 100 | 100 | 100 | Cortical dysplasia, complex, with other brain malformations 4, 615412 |
| TUBGCP2 | 99,1 | 95,5 | 97 | 97 | Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 |
| TUBGCP4 | 98,9 | 94,7 | 100 | 99,8 | Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 |
| TUBGCP6 | 100 | 99,4 | 100 | 100 | Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 |
| TUSC3 | 100 | 99,7 | 100 | 100 | Mental retardation, autosomal recessive 7, 611093 |
| TWIST1 | 100 | 99,4 | 96,7 | 90,6 | Craniosynostosis 1, 123100<br>Robinow-Sorauf syndrome, 180750<br>Sweeney-Cox syndrome, 617746<br>Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 |
| TWNK | 100 | 99,9 | 100 | 100 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245<br>Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286<br>Perrault syndrome 5, 616138 |
| U2AF2 | 99,8 | 97,7 | 100 | 100 | No OMIM disease ID |
| UBA5 | 97,4 | 86,6 | 100 | 100 | ?Spinocerebellar ataxia, autosomal recessive 24, 617133<br>Developmental and epileptic encephalopathy 44, 617132 |
| UBE2A | 99,5 | 97,4 | 100 | 100 | Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 |
| UBE3A | 98,9 | 94,1 | 100 | 100 | Angelman syndrome, 105830 |
| UBE3B | 100 | 99,7 | 100 | 100 | Kaufman oculocerebrofacial syndrome, 244450 |
| UBE4A | 99,7 | 98,7 | 100 | 100 | No OMIM disease ID |
| UBR1 | 99,6 | 99,1 | 98 | 97,9 | Johanson-Blizzard syndrome, 243800 |
| UBR7 | 99,9 | 99,9 | 100 | 100 | Li-Campeau syndrome, 619189 |
| UBTF | 99,9 | 99,1 | 100 | 100 | Neurodegeneration, childhood-onset, with brain atrophy, 617672 |
| UFC1 | 100 | 100 | 100 | 100 | Neurodevelopmental disorder with spasticity and poor growth, 618076 |
| UFM1 | 72,4 | 69,1 | 100 | 100 | Leukodystrophy, hypomyelinating, 14, 617899 |
| UFSP2 | 99,7 | 98,9 | 100 | 99,9 | ?Hip dysplasia, Beukes type, 142669<br>?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 |
| UGDH | 99,7 | 99,1 | 100 | 100 | Developmental and epileptic encephalopathy 84, 618792 |

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|---|---|---|---|---|---|
| UGP2 | 98,7 | 98,2 | 96,4 | 96,3 | Developmental and epileptic encephalopathy 83, 618744 |
| UNC13A | 99,4 | 97,8 | 100 | 99,9 | No OMIM disease ID |
| UNC80 | 97,9 | 97,1 | 100 | 100 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 |
| UPB1 | 100 | 100 | 100 | 100 | Beta-ureidopropionase deficiency, 613161 |
| UPF1 | 99,8 | 99,6 | 98,3 | 97,6 | No OMIM disease ID |
| UPF3B | 90,8 | 80,3 | 100 | 99,4 | Intellectual developmental disorder, X-linked syndromic 14, 300676 |
| UROC1 | 100 | 99,9 | 100 | 100 | ?Urocanase deficiency, 276880 |
| USP27X | 100 | 99,9 | 100 | 100 | Intellectual developmental disorder, X-linked 105, 300984 |
| USP7 | 90,8 | 85,2 | 94,8 | 94,7 | Hao-Fountain syndrome, 616863 |
| USP9X | 98,1 | 91,7 | 100 | 99,8 | Intellectual developmental disorder, X-linked 99, 300919<br>Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 |
| VAMP1 | 100 | 99,8 | 100 | 100 | Myasthenic syndrome, congenital, 25, 618323<br>Spastic ataxia 1, autosomal dominant, 108600 |
| VAMP2 | 99,1 | 97 | 100 | 100 | Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 |
| VARS1 | 100 | 99,7 | 100 | 100 | Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 |
| VARS2 | 100 | 99 | 100 | 100 | Combined oxidative phosphorylation deficiency 20, 615917 |
| VLDLR | 100 | 100 | 100 | 100 | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 |
| VPS11 | 94,6 | 92,2 | 100 | 100 | Leukodystrophy, hypomyelinating, 12, 616683 |
| VPS13B | 99,4 | 97,8 | 99,4 | 99,3 | Cohen syndrome, 216550 |
| VPS16 | 100 | 100 | 100 | 100 | Dystonia 30, 619291 |
| VPS35L | 100 | 99,8 | 100 | 100 | Ritscher-Schinzel syndrome 3, 619135 |
| VPS37A | 91,3 | 76 | 100 | 100 | Spastic paraplegia 53, autosomal recessive, 614898 |
| VPS41 | 99,8 | 98,8 | 100 | 100 | Spinocerebellar ataxia, autosomal recessive 29, 619389 |
| VPS4A | 100 | 99,9 | 100 | 100 | CIMDAG syndrome, 619273 |
| VPS53 | 91,1 | 89,9 | 100 | 99,4 | Pontocerebellar hypoplasia, type 2E, 615851 |
| VRK1 | 99,4 | 97,8 | 100 | 100 | Pontocerebellar hypoplasia type 1A, 607596 |
| VWA3B | 99,9 | 98,9 | 100 | 100 | ?Spinocerebellar ataxia, autosomal recessive 22, 616948 |
| WAC | 99,8 | 99,6 | 100 | 100 | Desanto-Shinawi syndrome, 616708 |
| WARS2 | 100 | 99,8 | 100 | 99,8 | Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 |

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|---|---|---|---|---|---|
| WASF1 | 99,2 | 94,1 | 100 | 100 | Neurodevelopmental disorder with absent language and variable seizures, 618707 |
| WASHC4 | 98,9 | 96 | 100 | 99,8 | ?Mental retardation, autosomal recessive 43, 615817 |
| WDFY3 | 99,8 | 99,1 | 100 | 100 | ?Microcephaly 18, primary, autosomal dominant, 617520 |
| WDPCP | 98 | 94,1 | 98,1 | 98 | ?Bardet-Biedl syndrome 15, 615992<br>Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 |
| WDR13 | 99,8 | 98,7 | 100 | 100 | No OMIM disease ID |
| WDR26 | 89,2 | 84,2 | 95,9 | 92,9 | Skraban-Deardorff syndrome, 617616 |
| WDR37 | 86,5 | 86,3 | 86,5 | 86,5 | Neurooculocardiogenitourinary syndrome, 618652 |
| WDR4 | 100 | 100 | 100 | 100 | Galloway-Mowat syndrome 6, 618347<br>Microcephaly, growth deficiency, seizures, and brain malformations, 618346 |
| WDR45 | 98,2 | 92 | 100 | 100 | Neurodegeneration with brain iron accumulation 5, 300894 |
| WDR45B | 94,8 | 80,3 | 100 | 100 | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 |
| WDR62 | 100 | 99,9 | 100 | 100 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 |
| WDR73 | 100 | 100 | 100 | 100 | Galloway-Mowat syndrome 1, 251300 |
| WDR81 | 100 | 100 | 100 | 100 | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185<br>Hydrocephalus, congenital, 3, with brain anomalies, 617967 |
| WFS1 | 100 | 99,8 | 100 | 100 | Deafness, autosomal dominant 6/14/38, 600965<br>?Cataract 41, 116400<br>Wolfram-like syndrome, autosomal dominant, 614296<br>Wolfram syndrome 1, 222300 |
| WWOX | 100 | 99,9 | 100 | 100 | Esophageal squamous cell carcinoma, somatic, 133239<br>Developmental and epileptic encephalopathy 28, 616211<br>Spinocerebellar ataxia, autosomal recessive 12, 614322 |
| XPA | 99,2 | 97,3 | 100 | 100 | Xeroderma pigmentosum, group A, 278700 |
| XRCC4 | 99,7 | 98,4 | 100 | 100 | Short stature, microcephaly, and endocrine dysfunction, 616541 |
| XYLT1 | 97,8 | 91,1 | 97,7 | 94,1 | Desbuquois dysplasia 2, 615777 |
| YIF1B | 99,9 | 99,2 | 90,1 | 90,1 | Kaya-Barakat-Masson syndrome, 619125 |
| YIPF5 | 100 | 100 | 100 | 100 | Microcephaly, epilepsy, and diabetes syndrome 2, 619278 |
| YME1L1 | 98,9 | 93,7 | 100 | 100 | ?Optic atrophy 11, 617302 |
| YWHAE | 100 | 100 | 100 | 100 | No OMIM disease ID |
| YWHAG | 100 | 99,9 | 100 | 100 | Developmental and epileptic encephalopathy 56, 617665 |

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|---|---|---|---|---|---|
| YY1 | 99,9 | 99,3 | 100 | 100 | Gabriele-de Vries syndrome, 617557 |
| ZBTB11 | 99,9 | 99,3 | 100 | 100 | Intellectual developmental disorder, autosomal recessive 69, 618383 |
| ZBTB16 | 100 | 100 | 100 | 100 | Skeletal defects, genital hypoplasia, and mental retardation, 612447<br>Leukemia, acute promyelocytic, PL2F/RARA type, |
| ZBTB18 | 100 | 99,8 | 100 | 99,8 | Mental retardation, autosomal dominant 22, 612337 |
| ZBTB20 | 100 | 100 | 100 | 100 | Primrose syndrome, 259050 |
| ZBTB24 | 100 | 100 | 100 | 100 | Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 |
| ZC3H14 | 99,7 | 98,2 | 100 | 99,9 | Mental retardation, autosomal recessive 56, 617125 |
| ZC4H2 | 100 | 98,1 | 100 | 100 | Wieacker-Wolff syndrome, 314580<br>Wieacker-Wolff syndrome, female-restricted, 301041 |
| ZDHHC9 | 97,3 | 84,5 | 100 | 100 | Mental retardation, X-linked syndromic, Raymond type, 300799 |
| ZEB2 | 99,7 | 98,5 | 97,4 | 97,4 | Mowat-Wilson syndrome, 235730 |
| ZFHX4 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| ZFYVE26 | 99,7 | 97,8 | 100 | 100 | Spastic paraplegia 15, autosomal recessive, 270700 |
| ZIC1 | 100 | 100 | 100 | 100 | ?Craniosynostosis 6, 616602<br>Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 |
| ZIC2 | 100 | 99,3 | 97,7 | 94,2 | Holoprosencephaly 5, 609637 |
| ZMIZ1 | 99,8 | 99 | 100 | 100 | Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 |
| ZMYM2 | 99,7 | 97,8 | 100 | 100 | No OMIM disease ID |
| ZMYND11 | 99,9 | 99,7 | 100 | 100 | Mental retardation, autosomal dominant 30, 616083 |
| ZNF142 | 100 | 99,7 | 100 | 100 | Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 |
| ZNF148 | 99,9 | 99,8 | 100 | 100 | Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 |
| ZNF292 | 99,5 | 98,1 | 99,6 | 99,6 | Intellectual developmental disorder, autosomal dominant 64, 619188 |
| ZNF335 | 100 | 99,7 | 100 | 100 | Microcephaly 10, primary, autosomal recessive, 615095 |
| ZNF407 | 99,9 | 99,1 | 100 | 100 | No OMIM disease ID |
| ZNF41 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| ZNF462 | 100 | 99,8 | 100 | 100 | Weiss-Kruszka syndrome, 618619 |
| ZNF526 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| ZNF699 | 99,9 | 99,2 | 100 | 100 | DEGCAGS syndrome, 619488 |
| ZNF711 | 99,4 | 96,5 | 100 | 99,8 | Intellectual developmental disorder, X-linked 97, 300803 |

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|---|---|---|---|---|---|
| ZSWIM6 | 95,1 | 91,6 | 94,3 | 91 | Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865<br>Acromelic frontonasal dysostosis, 603671 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*
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*Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.*
*TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : September 16th , 2021.*
*This list is accurate for panel version DG 3.2.0*
*Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors*