

# INTELLECTUAL DISABILITY GENE PANEL DG 2.18 (1338 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AAAS	100%	99,90%	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AASS	100%	99,70%	100%	100%	Hyperlysinemia, 238700
ABAT	100%	99,40%	100%	100%	GABA-transaminase deficiency, 613163
ABCC8	100%	99,80%	100%	100%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	100%	99,90%	100%	100%	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 10, 608569
ABCD1	75,80%	71,60%	100%	100%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD4	99,90%	98,60%	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	100%	100%	100%	100%	Chanarin-Dorfman syndrome, 275630
ACAD9	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	99,90%	98,20%	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACAT1	99,90%	97,50%	100%	100%	Alpha-methylacetoacetic aciduria, 203750
ACO2	96,30%	90,30%	100%	100%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100%	99,90%	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	100%	99,90%	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	98,70%	94,60%	100%	100%	Mental retardation, X-linked 63, 300387
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	100%	100%	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTL6A	99,80%	98,70%	100%	100%	No OMIM disease ID

<i>ACTL6B</i>	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
<i>ACVR1</i>	100%	100%	100%	100%	Fibrodysplasia ossificans progressiva, 135100
<i>ACY1</i>	100%	98,80%	100%	100%	Aminoacylase 1 deficiency, 609924
<i>ADAM22</i>	99,90%	99,50%	100%	100%	?Epileptic encephalopathy, early infantile, 61, 617933
<i>ADAR</i>	100%	99,80%	100%	100%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
<i>ADAT3</i>	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 36, 615286
<i>ADGRG1</i>	100%	100%	100%	100%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
<i>ADK</i>	99,50%	95,80%	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
<i>ADNP</i>	100%	100%	100%	100%	Helsmoortel-van der Aa syndrome, 615873
<i>ADPRHL2</i>	100%	99,80%	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
<i>ADSL</i>	99,20%	98,70%	100%	100%	Adenylosuccinase deficiency, 103050
<i>AFF2</i>	99,90%	99,40%	100%	99,80%	Mental retardation, X-linked, FRAXE type, 309548
<i>AFF4</i>	99,90%	98,90%	100%	100%	CHOPS syndrome, 616368
<i>AFG3L2</i>	95,00%	91,10%	100%	99,90%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
<i>AGA</i>	100%	100%	100%	100%	Aspartylglucosaminuria, 208400
<i>AGMO</i>	99,20%	96,00%	100%	100%	No OMIM disease ID
<i>AGO2</i>	99,10%	99,10%	99,90%	99,50%	No OMIM disease ID
<i>AHCY</i>	100%	99,20%	100%	100%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
<i>AHDC1</i>	100%	99,30%	100%	100%	Xia-Gibbs syndrome, 615829
<i>AHI1</i>	99,70%	97,90%	100%	100%	Joubert syndrome 3, 608629
<i>AHSG</i>	99,90%	99,50%	100%	100%	?Alopecia-mental retardation syndrome 1, 203650
<i>AIFM1</i>	99,90%	98,80%	100%	100%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
<i>AIMP1</i>	99,20%	94,50%	100%	99,90%	Leukodystrophy, hypomyelinating, 3, 260600
<i>AIMP2</i>	88,90%	86,00%	100%	100%	Leukodystrophy, hypomyelinating, 17, 618006
<i>AKT3</i>	98,70%	94,50%	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
<i>ALDH18A1</i>	100%	99,90%	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
<i>ALDH3A2</i>	95,30%	94,60%	100%	100%	Sjogren-Larsson syndrome, 270200

<i>ALDH4A1</i>	100%	99,40%	100%	100%	Hyperprolinemia, type II, 239510
<i>ALDH5A1</i>	91,00%	81,50%	100%	100%	Succinic semialdehyde dehydrogenase deficiency, 271980
<i>ALDH7A1</i>	94,40%	88,80%	100%	100%	Epilepsy, pyridoxine-dependent, 266100
<i>ALG1</i>	53,00%	45,80%	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
<i>ALG11</i>	96,80%	96,80%	96,80%	96,80%	Congenital disorder of glycosylation, type Ip, 613661
<i>ALG12</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
<i>ALG13</i>	98,40%	92,60%	100%	99,60%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
<i>ALG2</i>	100%	100%	100%	100%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
<i>ALG3</i>	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type Id, 601110
<i>ALG6</i>	98,60%	94,80%	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
<i>ALG8</i>	97,20%	95,60%	96,60%	96,60%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
<i>ALG9</i>	100%	99,70%	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type II, 608776
<i>ALKBH8</i>	99,80%	98,90%	100%	100%	Intellectual developmental disorder, autosomal recessive 71, 618504
<i>ALMS1</i>	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
<i>ALX3</i>	77,90%	73,30%	100%	100%	Frontonasal dysplasia 1, 136760
<i>ALX4</i>	100%	99,30%	100%	100%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
<i>AMER1</i>	99,90%	98,50%	100%	100%	Osteopathia striata with cranial sclerosis, 300373
<i>AMMECR1</i>	100%	99,10%	100%	100%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
<i>AMPD2</i>	99,80%	98,90%	100%	100%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
<i>AMT</i>	100%	100%	100%	100%	Glycine encephalopathy, 605899
<i>ANK3</i>	99,30%	99,00%	100%	100%	?Mental retardation, autosomal recessive, 37, 615493
<i>ANKH</i>	100%	100%	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
<i>ANKLE2</i>	99,90%	98,60%	100%	99,80%	Microcephaly 16, primary, autosomal recessive, 616681
<i>ANKRD11</i>	97,50%	94,80%	100%	100%	KBG syndrome, 148050
<i>ANKS1B</i>	100%	99,60%	100%	100%	No OMIM disease ID
<i>ANO10</i>	99,80%	97,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
<i>ANTXR1</i>	99,70%	97,90%	100%	100%	GAP0 syndrome, 230740
<i>AP1S1</i>	99,90%	99,50%	100%	100%	MEDNIK syndrome, 609313
<i>AP1S2</i>	76,40%	67,90%	100%	100%	Mental retardation, X-linked syndromic 5, 304340
<i>AP2M1</i>	100%	100%	100%	100%	Intellectual developmental disorder 60 with seizures, 618587

AP3B1	99,20%	95,80%	100%	100%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	99,40%	95,10%	100%	100%	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	99,80%	98,60%	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	99,90%	98,70%	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	99,80%	98,70%	100%	100%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,90%	98,90%	100%	100%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,90%	71,30%	87,90%	87,90%	Spastic paraplegia 52, autosomal recessive, 614067
APC2	97,60%	92,70%	99,90%	99,10%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APOPT1	81,90%	80,70%	93,50%	93,40%	Mitochondrial complex IV deficiency, 220110
APTX	94,90%	92,50%	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	97,00%	96,60%	96,90%	96,60%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARFGEF2	99,90%	99,10%	100%	100%	Periventricular heterotopia with microcephaly, 608097
ARG1	100%	100%	100%	100%	Argininemia, 207800
ARHGAP31	99,90%	98,80%	100%	100%	Adams-Oliver syndrome 1, 100300
ARHGEF6	99,50%	96,20%	100%	99,90%	No OMIM disease ID
ARHGEF9	76,50%	74,10%	97,20%	97,10%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	98,10%	96,40%	100%	100%	Coffin-Siris syndrome 2, 614607
ARID1B	99,50%	98,60%	99,90%	99,20%	Coffin-Siris syndrome 1, 135900
ARID2	99,80%	98,40%	100%	100%	Coffin-Siris syndrome 6, 617808
ARL13B	100%	99,20%	100%	100%	Joubert syndrome 8, 612291
ARL6	99,90%	98,60%	100%	100%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC9	100%	99,80%	100%	100%	Joubert syndrome 30, 617622
ARSA	100%	99,80%	100%	100%	Metachromatic leukodystrophy, 250100
ARSE	99,00%	93,00%	100%	99,90%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	81,00%	64,00%	91,50%	85,70%	Proud syndrome, 300004 Partington syndrome, 309510 Lissencephaly, X-linked 2, 300215 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	99,70%	98,60%	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASH1L	98,70%	98,60%	98,70%	98,70%	Mental retardation, autosomal dominant 52, 617796

ASL	100%	99,60%	100%	100%	Argininosuccinic aciduria, 207900
ASNS	99,40%	95,20%	100%	100%	Asparagine synthetase deficiency, 615574
ASPA	99,90%	98,30%	100%	100%	Canavan disease, 271900
ASPM	99,70%	98,20%	100%	100%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	95,40%	87,90%	100%	100%	Citrullinemia, 215700
ASXL1	100%	99,50%	99,90%	99,90%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	99,70%	98,90%	100%	100%	Shashi-Pena syndrome, 617190
ASXL3	99,90%	99,70%	100%	100%	Bainbridge-Ropers syndrome, 615485
ATAD1	99,60%	95,10%	100%	100%	Hyperekplexia 4, 618011
ATAD3A	91,90%	83,20%	100%	100%	Harel-Yoon syndrome, 617183 ?Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATIC	99,90%	99,30%	100%	100%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100%	99,70%	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATN1	99,90%	98,20%	100%	100%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP1A1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	100%	100%	100%	100%	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	100%	99,90%	100%	100%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2A2	100%	100%	100%	100%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP2	94,10%	76,60%	100%	100%	Congenital disorder of glycosylation, type IIr, 301045 Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP6VOA2	100%	99,50%	100%	100%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	99,90%	98,70%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 3, 618012 Cutis laxa, autosomal recessive, type IID, 617403
ATP6V1B2	100%	99,30%	100%	100%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480

<i>ATP7A</i>	99,70%	97,50%	100%	100%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
<i>ATP8A2</i>	100%	99,70%	100%	100%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
<i>ATR</i>	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
<i>ATRX</i>	99,40%	96,30%	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
<i>AUH</i>	100%	99,80%	100%	100%	3-methylglutaconic aciduria, type I, 250950
<i>AUTS2</i>	98,20%	95,80%	100%	100%	Mental retardation, autosomal dominant 26, 615834
<i>AVPR2</i>	100%	99,40%	100%	100%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
<i>B3GALNT2</i>	93,80%	89,40%	92,50%	92,50%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
<i>B3GALT6</i>	75,70%	69,70%	89,80%	81,60%	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
<i>B3GLCT</i>	99,60%	96,30%	99,90%	99,20%	Peters-plus syndrome, 261540
<i>B4GALNT1</i>	99,30%	95,00%	100%	100%	Spastic paraplegia 26, autosomal recessive, 609195
<i>B4GALT7</i>	99,80%	97,40%	99,90%	98,60%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
<i>B4GAT1</i>	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
<i>BAZ2B</i>	99,90%	99,00%	100%	100%	No OMIM disease ID
<i>BBS1</i>	100%	100%	100%	100%	Bardet-Biedl syndrome 1, 209900
<i>BBS10</i>	100%	99,80%	100%	100%	Bardet-Biedl syndrome 10, 615987
<i>BBS12</i>	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
<i>BBS2</i>	100%	99,50%	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
<i>BBS4</i>	99,90%	99,30%	100%	100%	Bardet-Biedl syndrome 4, 615982
<i>BBS5</i>	99,00%	93,90%	100%	100%	Bardet-Biedl syndrome 5, 615983
<i>BBS7</i>	98,70%	95,50%	100%	100%	Bardet-Biedl syndrome 7, 615984
<i>BBS9</i>	99,70%	97,60%	100%	100%	Bardet-Biedl syndrome 9, 615986
<i>BCAP31</i>	92,60%	83,20%	100%	99,90%	Deafness, dystonia, and cerebral hypomyelination, 300475
<i>BCKDHA</i>	99,90%	99,20%	100%	100%	Maple syrup urine disease, type Ia, 248600
<i>BCKDHB</i>	99,50%	94,40%	100%	100%	Maple syrup urine disease, type Ib, 248600
<i>BCKDK</i>	100%	100%	100%	100%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
<i>BCL11A</i>	99,80%	98,10%	100%	100%	Dias-Logan syndrome, 617101

<i>BCL11B</i>	99,10%	95,60%	98,80%	97,30%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
<i>BCOR</i>	99,60%	97,40%	100%	99,90%	Microphthalmia, syndromic 2, 300166
<i>BCORL1</i>	99,60%	97,90%	100%	100%	Shukla-Vernon syndrome, 301029
<i>BCS1L</i>	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
<i>BLM</i>	99,80%	98,30%	100%	100%	Bloom syndrome, 210900
<i>BOLA3</i>	99,40%	90,20%	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
<i>BPTF</i>	96,10%	94,20%	99,60%	98,60%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
<i>BRAF</i>	95,60%	85,10%	100%	100%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
<i>BRAT1</i>	99,70%	98,20%	100%	100%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
<i>BRF1</i>	99,90%	98,40%	100%	100%	Cerebellofaciodental syndrome, 616202
<i>BRPF1</i>	100%	100%	100%	100%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
<i>BRSK2</i>	99,50%	97,40%	100%	100%	No OMIM disease ID
<i>BRWD3</i>	99,30%	97,20%	100%	100%	Mental retardation, X-linked 93, 300659
<i>BSCL2</i>	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
<i>BTB</i>	100%	99,90%	100%	100%	Biotinidase deficiency, 253260
<i>BUB1B</i>	99,60%	98,90%	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
<i>C12orf4</i>	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 66, 618221
<i>C12orf57</i>	100%	98,90%	100%	100%	Temtamy syndrome, 218340
<i>C12orf65</i>	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
<i>C2CD3</i>	95,80%	95,60%	95,90%	95,90%	Orofaciodigital syndrome XIV, 615948
<i>C5orf42</i>	99,70%	98,40%	100%	100%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170

CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	99,70%	97,10%	100%	100%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	99,60%	97,30%	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	95,10%	91,90%	100%	100%	Spinocerebellar ataxia 6, 183086 Epileptic encephalopathy, early infantile, 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	97,50%	95,70%	99,10%	97,70%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	99,90%	99,20%	100%	100%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	98,00%	97,90%	100%	100%	Sinoatrial node dysfunction and deafness, 614896 Primary aldosteronism, seizures, and neurologic abnormalities, 615474
CACNA1E	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1G	100%	99,60%	100%	100%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA2D2	94,00%	93,20%	99,20%	97,60%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 50, 616457
CAMK2A	100%	99,60%	100%	100%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	100%	99,80%	100%	100%	Mental retardation, autosomal dominant 54, 617799
CAMK2G	99,90%	98,10%	100%	100%	Mental retardation, autosomal dominant 59, 618522
CAMTA1	100%	99,50%	100%	100%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	100%	99,90%	100%	100%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CARS2	100%	100%	100%	99,20%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	99,60%	96,80%	100%	100%	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CBL	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	99,80%	98,30%	100%	100%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360



<i>CCBE1</i>	99,80%	98,80%	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
<i>CCDC115</i>	95,30%	90,00%	100%	100%	Congenital disorder of glycosylation, type Ilo, 616828
<i>CCDC174</i>	99,50%	97,10%	100%	100%	Hypotonia, infantile, with psychomotor retardation, 616816
<i>CCDC22</i>	99,60%	96,50%	100%	100%	Ritscher-Schinzel syndrome 2, 300963
<i>CCDC47</i>	99,40%	97,50%	100%	100%	Trichohepatoneurodevelopmental syndrome, 618268
<i>CCDC88A</i>	98,90%	95,40%	100%	100%	?PEHO syndrome-like, 617507
<i>CCDC88C</i>	100%	99,30%	100%	100%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
<i>CCND2</i>	100%	100%	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
<i>CCNK</i>	92,60%	89,80%	100%	99,80%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
<i>CDC42</i>	97,80%	90,70%	100%	100%	Takenouchi-Kosaki syndrome, 616737
<i>CDC42BPB</i>	100%	99,30%	100%	100%	No OMIM disease ID
<i>CDC6</i>	100%	100%	100%	100%	?Meier-Gorlin syndrome 5, 613805
<i>CDH11</i>	100%	100%	100%	100%	Elsahy-Waters syndrome, 211380
<i>CDH15</i>	99,90%	98,70%	100%	100%	Mental retardation, autosomal dominant 3, 612580
<i>CDK10</i>	100%	99,90%	100%	100%	Al Kaissi syndrome, 617694
<i>CDK13</i>	98,00%	92,70%	100%	100%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
<i>CDK5RAP2</i>	99,80%	98,90%	100%	100%	Microcephaly 3, primary, autosomal recessive, 604804
<i>CDK8</i>	99,70%	97,90%	100%	100%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
<i>CDKL5</i>	95,00%	93,50%	95,60%	95,00%	Epileptic encephalopathy, early infantile, 2, 300672
<i>CDKN1C</i>	86,30%	74,80%	99,20%	96,90%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
<i>CDON</i>	100%	99,60%	100%	100%	Holoprosencephaly 11, 614226
<i>CENPF</i>	99,80%	98,50%	100%	100%	Stromme syndrome, 243605
<i>CENPJ</i>	100%	99,60%	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
<i>CEP104</i>	100%	99,20%	100%	100%	Joubert syndrome 25, 616781
<i>CEP120</i>	100%	99,50%	100%	100%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
<i>CEP135</i>	99,10%	93,60%	100%	100%	Microcephaly 8, primary, autosomal recessive, 614673
<i>CEP152</i>	99,70%	98,20%	100%	100%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
<i>CEP290</i>	96,10%	90,00%	100%	100%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188

<i>CEP41</i>	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
<i>CEP57</i>	99,20%	93,00%	100%	100%	Mosaic variegated aneuploidy syndrome 2, 614114
<i>CEP83</i>	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
<i>CEP89</i>	96,00%	94,50%	100%	100%	No OMIM disease ID
<i>CHAMP1</i>	100%	100%	100%	100%	Mental retardation, autosomal dominant 40, 616579
<i>CHD1</i>	99,30%	94,90%	100%	100%	Pilarowski-Bjornsson syndrome, 617682
<i>CHD2</i>	99,40%	99,20%	100%	100%	Epileptic encephalopathy, childhood-onset, 615369
<i>CHD3</i>	94,80%	92,60%	99,80%	99,50%	Snijders Blok-Campeau syndrome, 618205
<i>CHD4</i>	100%	99,90%	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
<i>CHD7</i>	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
<i>CHD8</i>	100%	99,90%	100%	100%	No OMIM disease ID
<i>CHKB</i>	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
<i>CHMP1A</i>	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
<i>CHRNA4</i>	98,30%	96,20%	100%	100%	Epilepsy, nocturnal frontal lobe, 1, 600513
<i>CIC</i>	63,30%	63,30%	100%	99,90%	Mental retardation, autosomal dominant 45, 617600
<i>CIT</i>	100%	99,40%	100%	100%	Microcephaly 17, primary, autosomal recessive, 617090
<i>CKAP2L</i>	99,70%	98,60%	100%	100%	Filippi syndrome, 272440
<i>CLCN4</i>	99,90%	98,90%	100%	100%	Raynaud-Claes syndrome, 300114
<i>CLIC2</i>	99,90%	96,50%	100%	100%	?Mental retardation, X-linked, syndromic 32, 300886
<i>CLIP1</i>	100%	99,00%	100%	100%	No OMIM disease ID
<i>CLN3</i>	92,50%	91,80%	92,50%	92,50%	Ceroid lipofuscinosis, neuronal, 3, 204200
<i>CLN5</i>	99,40%	95,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
<i>CLN6</i>	99,90%	97,10%	100%	100%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
<i>CLN8</i>	83,50%	83,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
<i>CLP1</i>	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 10, 615803
<i>CLPB</i>	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
<i>CLTC</i>	100%	99,90%	100%	100%	Mental retardation, autosomal dominant 56, 617854
<i>CNKS2</i>	98,60%	93,60%	100%	100%	Mental retardation, X-linked, syndromic, Houge type, 301008
<i>CNNM2</i>	100%	100%	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
<i>CNOT1</i>	100%	99,90%	100%	100%	Holoprosencephaly 12, with or without pancreatic agenesis, 618500
<i>CNOT2</i>	99,90%	99,50%	100%	100%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
<i>CNOT3</i>	100%	100%	100%	100%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672

<i>CNPY3</i>	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 60, 617929
<i>CNTNAP2</i>	100%	99,80%	100%	100%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
<i>COASY</i>	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
<i>COG1</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIg, 611209
<i>COG4</i>	100%	99,90%	100%	100%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
<i>COG5</i>	99,70%	97,60%	100%	100%	Congenital disorder of glycosylation, type IIi, 613612
<i>COG6</i>	99,10%	93,90%	100%	100%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
<i>COG7</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
<i>COG8</i>	100%	99,60%	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
<i>COL4A1</i>	98,70%	97,40%	100%	100%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
<i>COL4A2</i>	100%	99,60%	100%	100%	Brain small vessel disease 2, 614483
<i>COL4A3BP</i>	99,60%	96,50%	100%	100%	Mental retardation, autosomal dominant 34, 616351
<i>COLEC11</i>	100%	100%	100%	100%	3MC syndrome 2, 265050
<i>COQ2</i>	98,00%	95,30%	97,20%	97,20%	Coenzyme Q10 deficiency, primary, 1, 607426
<i>COQ4</i>	90,90%	89,30%	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
<i>COQ8A</i>	100%	99,50%	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016
<i>COQ9</i>	100%	97,90%	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
<i>COX10</i>	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
<i>COX15</i>	99,90%	98,80%	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
<i>COX6B1</i>	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110
<i>CPLX1</i>	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 63, 617976
<i>CPS1</i>	100%	99,90%	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300
<i>CRADD</i>	99,50%	96,30%	100%	100%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
<i>CRBN</i>	88,20%	87,70%	97,00%	92,90%	Mental retardation, autosomal recessive 2, 607417
<i>CREBBP</i>	99,70%	98,50%	100%	100%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
<i>CRLF1</i>	91,00%	89,80%	97,90%	95,20%	Cold-induced sweating syndrome 1, 272430
<i>CSDE1</i>	99,90%	99,50%	100%	100%	No OMIM disease ID

<i>CSF1R</i>	99,90%	99,30%	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
<i>CSNK2A1</i>	95,90%	92,20%	92,30%	92,30%	Okur-Chung neurodevelopmental syndrome, 617062
<i>CSNK2B</i>	100%	100%	100%	100%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
<i>CSPP1</i>	99,80%	98,70%	100%	100%	Joubert syndrome 21, 615636
<i>CSTB</i>	99,60%	89,80%	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
<i>CTBP1</i>	93,20%	86,90%	99,50%	98,60%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
<i>CTC1</i>	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
<i>CTCF</i>	100%	99,30%	100%	100%	Mental retardation, autosomal dominant 21, 615502
<i>CTDP1</i>	88,40%	84,30%	100%	99,40%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
<i>CTNNA2</i>	100%	99,80%	100%	100%	Cortical dysplasia, complex, with other brain malformations 9, 618174
<i>CTNNB1</i>	100%	100%	100%	100%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
<i>CTNND2</i>	93,50%	91,10%	97,70%	95,50%	No OMIM disease ID
<i>CTSA</i>	100%	100%	100%	100%	Galactosialidosis, 256540
<i>CTSD</i>	98,40%	95,00%	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
<i>CTTNBP2</i>	99,50%	97,30%	100%	100%	No OMIM disease ID
<i>CTU2</i>	99,70%	97,70%	100%	100%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
<i>CUL4B</i>	98,00%	90,90%	99,90%	99,20%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
<i>CUX1</i>	96,40%	94,80%	99,30%	98,40%	Global developmental delay with or without impaired intellectual development, 618330
<i>CUX2</i>	99,90%	99,10%	100%	100%	Epileptic encephalopathy, early infantile, 67, 618141
<i>CWC27</i>	99,30%	96,50%	100%	100%	Retinitis pigmentosa with or without skeletal anomalies, 250410
<i>CWF19L1</i>	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive 17, 616127
<i>CXorf56</i>	99,80%	96,70%	100%	100%	?Mental retardation, X-linked 107, 301013
<i>CYB5R3</i>	98,40%	98,00%	99,80%	98,90%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
<i>CYFIP2</i>	100%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 65, 618008
<i>CYP27A1</i>	98,90%	96,70%	100%	100%	Cerebrotendinous xanthomatosis, 213700
<i>CYP2U1</i>	94,80%	91,50%	100%	99,90%	Spastic paraplegia 56, autosomal recessive, 615030
<i>D2HGDH</i>	99,20%	97,20%	100%	100%	D-2-hydroxyglutaric aciduria, 600721
<i>DAG1</i>	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538

<i>DARS</i>	100%	99,30%	100%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
<i>DARS2</i>	100%	99,30%	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
<i>DBT</i>	99,80%	98,00%	100%	100%	Maple syrup urine disease, type II, 248600
<i>DCAF17</i>	98,90%	93,30%	100%	100%	Woodhouse-Sakati syndrome, 241080
<i>DCC</i>	100%	100%	100%	100%	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500
<i>DCHS1</i>	99,80%	99,10%	100%	100%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
<i>DCPS</i>	100%	100%	100%	100%	Al-Raqad syndrome, 616459
<i>DCX</i>	100%	99,90%	100%	100%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
<i>DDC</i>	99,70%	96,40%	100%	100%	Aromatic L-amino acid decarboxylase deficiency, 608643
<i>DDHD2</i>	100%	99,60%	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
<i>DDX11</i>	85,20%	80,70%	100%	100%	Warsaw breakage syndrome, 613398
<i>DDX3X</i>	86,80%	84,90%	100%	100%	Mental retardation, X-linked 102, 300958
<i>DDX59</i>	100%	100%	100%	100%	Orofaciodigital syndrome V, 174300
<i>DDX6</i>	97,70%	88,70%	100%	100%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
<i>DEAF1</i>	97,30%	88,80%	100%	98,70%	Mental retardation, autosomal dominant 24, 615828 ?Dyskinesia, seizures, and intellectual developmental disorder, 617171
<i>DEGS1</i>	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 18, 618404
<i>DENND5A</i>	100%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 49, 617281
<i>DEPDC5</i>	100%	99,80%	100%	100%	Epilepsy, familial focal, with variable foci 1, 604364
<i>DHCR24</i>	100%	100%	100%	100%	Desmosterolosis, 602398
<i>DHCR7</i>	100%	100%	100%	100%	Smith-Lemli-Opitz syndrome, 270400
<i>DHDDS</i>	99,00%	95,00%	95,20%	95,20%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
<i>DHFR</i>	92,10%	78,90%	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
<i>DHPS</i>	100%	99,70%	93,30%	93,20%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
<i>DHTKD1</i>	99,90%	98,90%	100%	100%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
<i>DHX30</i>	100%	99,90%	100%	100%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
<i>DIAPH1</i>	99,80%	99,00%	99,50%	97,90%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
<i>DIP2B</i>	100%	99,30%	100%	100%	Mental retardation, FRA12A type, 136630

<i>DIS3L2</i>	100%	99,80%	100%	100%	Perlman syndrome, 267000
<i>DKC1</i>	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
<i>DLD</i>	100%	99,70%	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
<i>DLG3</i>	99,10%	93,20%	100%	100%	Mental retardation, X-linked 90, 300850
<i>DLG4</i>	100%	99,90%	100%	100%	Intellectual developmental disorder 62, 618793
<i>DMD</i>	99,60%	98,60%	100%	100%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
<i>DMPK</i>	99,80%	98,40%	100%	100%	Myotonic dystrophy 1, 160900
<i>DMXL2</i>	99,90%	99,10%	100%	100%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113 Epileptic encephalopathy, early infantile, 81, 618663
<i>DNAJC12</i>	87,40%	87,40%	100%	100%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
<i>DNAJC19</i>	98,90%	96,20%	100%	100%	3-methylglutaconic aciduria, type V, 610198
<i>DNM1</i>	92,60%	89,10%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 31, 616346
<i>DNMT3A</i>	99,80%	98,60%	100%	100%	Heyn-Sproul-Jackson syndrome, 618724 Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
<i>DNMT3B</i>	100%	100%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
<i>DOCK3</i>	100%	99,00%	100%	100%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
<i>DOCK6</i>	99,30%	98,90%	100%	100%	Adams-Oliver syndrome 2, 614219
<i>DOCK7</i>	99,80%	98,20%	100%	99,90%	Epileptic encephalopathy, early infantile, 23, 615859
<i>DOLK</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type Im, 610768
<i>DONSON</i>	91,70%	85,30%	100%	100%	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
<i>DPAGT1</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
<i>DPF2</i>	99,90%	98,40%	100%	100%	Coffin-Siris syndrome 7, 618027
<i>DPH1</i>	100%	99,90%	100%	100%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
<i>DPM1</i>	98,20%	91,30%	99,70%	97,10%	Congenital disorder of glycosylation, type Ie, 608799
<i>DPP6</i>	99,70%	97,80%	99,40%	97,60%	Mental retardation, autosomal dominant 33, 616311
<i>DPYD</i>	99,70%	97,70%	100%	100%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
<i>DPYS</i>	100%	99,90%	100%	100%	Dihydropyrimidinuria, 222748
<i>DYM</i>	97,40%	96,50%	100%	100%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800

<i>DYNC1H1</i>	99,90%	99,40%	100%	100%	Mental retardation, autosomal dominant 13, 614563 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
<i>DYNC1I2</i>	84,40%	68,80%	100%	100%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
<i>DYRK1A</i>	100%	100%	100%	100%	Mental retardation, autosomal dominant 7, 614104
<i>EBF3</i>	100%	100%	100%	100%	Hypotonia, ataxia, and delayed development syndrome, 617330
<i>EBP</i>	99,70%	95,80%	100%	100%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
<i>ECHS1</i>	99,90%	99,00%	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
<i>EDC3</i>	100%	99,90%	100%	100%	?Mental retardation, autosomal recessive 50, 616460
<i>EED</i>	99,10%	95,90%	100%	100%	Cohen-Gibson syndrome, 617561
<i>EEF1A2</i>	100%	100%	99,90%	99,10%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
<i>EFNB2</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>EFTUD2</i>	100%	99,80%	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
<i>EHMT1</i>	94,50%	93,70%	99,60%	99,50%	Kleefstra syndrome 1, 610253
<i>EIF2AK3</i>	97,20%	94,50%	100%	100%	Wolcott-Rallison syndrome, 226980
<i>EIF2S3</i>	95,40%	89,10%	100%	100%	MEHMO syndrome, 300148
<i>EIF3F</i>	96,80%	84,10%	100%	100%	Mental retardation, autosomal recessive 67, 618295
<i>EIF4A3</i>	100%	99,50%	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
<i>ELAC2</i>	100%	99,70%	100%	100%	Combined oxidative phosphorylation deficiency 17, 615440
<i>ELOVL4</i>	100%	99,50%	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
<i>ELP2</i>	99,90%	98,80%	100%	100%	Mental retardation, autosomal recessive 58, 617270
<i>EMC1</i>	100%	99,30%	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
<i>EML1</i>	99,70%	98,40%	100%	100%	Band heterotopia, 600348
<i>EMX2</i>	100%	100%	100%	100%	Schizencephaly, 269160
<i>ENTPD1</i>	100%	100%	100%	100%	Spastic paraplegia 64, autosomal recessive, 615683
<i>EP300</i>	99,80%	99,00%	100%	100%	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
<i>EPG5</i>	99,50%	98,50%	100%	100%	Vici syndrome, 242840
<i>ERCC1</i>	100%	99,30%	100%	100%	Cerebrooculofacioskeletal syndrome 4, 610758
<i>ERCC2</i>	100%	99,70%	100%	100%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730

<i>ERCC3</i>	100%	99,40%	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
<i>ERCC5</i>	100%	99,60%	100%	100%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
<i>ERCC6</i>	100%	100%	100%	100%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
<i>ERCC8</i>	99,50%	95,80%	100%	100%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
<i>ERLIN2</i>	100%	99,90%	100%	100%	Spastic paraplegia 18, autosomal recessive, 611225
<i>ESCO2</i>	98,70%	95,20%	100%	100%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
<i>ETFB</i>	100%	99,80%	100%	100%	Glutaric acidemia IIB, 231680
<i>ETHE1</i>	99,90%	97,40%	100%	100%	Ethylmalonic encephalopathy, 602473
<i>EXOSC2</i>	100%	100%	100%	100%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
<i>EXOSC3</i>	99,50%	94,90%	100%	100%	Pontocerebellar hypoplasia, type 1B, 614678
<i>EXOSC9</i>	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 1D, 618065
<i>EXTL3</i>	100%	100%	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
<i>EZH2</i>	100%	99,50%	100%	100%	Weaver syndrome, 277590
<i>FA2H</i>	92,00%	83,10%	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
<i>FAM126A</i>	100%	99,40%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
<i>FAM20C</i>	100%	100%	100%	99,80%	Raine syndrome, 259775
<i>FAR1</i>	97,60%	92,80%	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
<i>FARS2</i>	100%	100%	100%	100%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
<i>FARSB</i>	98,80%	94,60%	100%	100%	Rajab interstitial lung disease with brain calcifications, 613658
<i>FAT4</i>	100%	100%	100%	100%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
<i>FBXL3</i>	100%	100%	100%	100%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
<i>FBXL4</i>	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
<i>FBXO11</i>	96,90%	92,70%	100%	100%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
<i>FBXO31</i>	96,00%	93,10%	100%	99,90%	?Mental retardation, autosomal recessive 45, 615979
<i>FDFT1</i>	97,70%	96,00%	100%	100%	Squalene synthase deficiency, 618156
<i>FGD1</i>	97,30%	92,80%	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400



<i>FGF12</i>	99,90%	98,10%	100%	100%	Epileptic encephalopathy, early infantile, 47, 617166
<i>FGF14</i>	100%	100%	100%	100%	Spinocerebellar ataxia 27, 609307
<i>FGFR1</i>	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 HEARTsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
<i>FGFR2</i>	97,70%	97,10%	100%	100%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
<i>FGFR3</i>	99,80%	97,70%	100%	99,80%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482

<i>FH</i>	92,10%	88,30%	100%	100%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
<i>FIBP</i>	100%	100%	100%	100%	Thauvin-Robinet-Faivre syndrome, 617107
<i>FIGN</i>	100%	100%	100%	100%	No OMIM disease ID
<i>FMR1</i>	96,10%	92,10%	100%	100%	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624
<i>FKRP</i>	100%	100%	100%	99,90%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
<i>FKTN</i>	99,70%	97,00%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
<i>FLNA</i>	100%	99,90%	100%	100%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
<i>FLVCR1</i>	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
<i>FLVCR2</i>	100%	100%	100%	100%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
<i>FMN2</i>	85,50%	82,50%	100%	100%	Mental retardation, autosomal recessive 47, 616193
<i>FOLR1</i>	100%	100%	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
<i>FOXG1</i>	88,60%	82,10%	99,20%	96,40%	Rett syndrome, congenital variant, 613454
<i>FOXJ1</i>	99,90%	98,40%	100%	100%	Ciliary dyskinesia, primary, 43, 618699
<i>FOXP1</i>	100%	99,80%	100%	100%	Mental retardation with language impairment and with or without autistic features, 613670
<i>FOXP2</i>	99,50%	99,20%	100%	100%	Speech-language disorder-1, 602081
<i>FOXRED1</i>	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 19, 618241
<i>FRAS1</i>	100%	99,40%	100%	100%	Fraser syndrome 1, 219000
<i>FRMD4A</i>	90,70%	87,30%	96,60%	96,60%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
<i>FRMPD4</i>	99,80%	98,50%	100%	100%	Mental retardation, X-linked 104, 300983
<i>FRRS1L</i>	79,70%	69,10%	99,20%	95,80%	Epileptic encephalopathy, early infantile, 37, 616981
<i>FTCD</i>	95,70%	91,00%	100%	100%	Glutamate formiminotransferase deficiency, 229100

<i>FTO</i>	83,80%	83,70%	94,20%	94,20%	Growth retardation, developmental delay, facial dysmorphism, 612938
<i>FTSJ1</i>	98,00%	93,80%	100%	100%	Mental retardation, X-linked 9/44, 309549
<i>FUCA1</i>	100%	99,90%	100%	100%	Fucosidosis, 230000
<i>FUT8</i>	100%	99,20%	100%	100%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
<i>GABBR2</i>	96,20%	92,00%	99,10%	98,40%	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 Epileptic encephalopathy, early infantile, 59, 617904
<i>GABRA1</i>	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744
<i>GABRA3</i>	98,70%	94,40%	99,90%	98,90%	No OMIM disease ID
<i>GABRB1</i>	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 45, 617153
<i>GABRB2</i>	100%	99,90%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
<i>GABRB3</i>	99,60%	98,20%	100%	100%	Epileptic encephalopathy, early infantile, 43, 617113
<i>GABRG2</i>	90,80%	90,20%	93,00%	93,00%	Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
<i>GAD1</i>	100%	99,90%	100%	100%	?Cerebral palsy, spastic quadriplegic, 1, 603513
<i>GALC</i>	99,80%	98,30%	100%	100%	Krabbe disease, 245200
<i>GALE</i>	100%	100%	100%	100%	Galactose epimerase deficiency, 230350
<i>GALT</i>	100%	99,70%	100%	100%	Galactosemia, 230400
<i>GAMT</i>	93,10%	82,70%	100%	100%	Cerebral creatine deficiency syndrome 2, 612736
<i>GATAD2B</i>	100%	100%	100%	100%	Mental retardation, autosomal dominant 18, 615074
<i>GATM</i>	100%	100%	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
<i>GCH1</i>	99,90%	95,50%	100%	100%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
<i>GCSH</i>	75,70%	68,90%	100%	100%	?Glycine encephalopathy, 605899
<i>GDI1</i>	99,80%	98,70%	100%	100%	Mental retardation, X-linked 41, 300849
<i>GFAP</i>	91,80%	89,70%	100%	100%	Alexander disease, 203450
<i>GFM1</i>	99,90%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
<i>GFM2</i>	98,90%	95,20%	100%	100%	Combined oxidative phosphorylation deficiency 39, 618397
<i>GJA1</i>	100%	100%	100%	100%	Erythrokeratoderma variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
<i>GJB1</i>	100%	100%	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800

<i>GJC2</i>	78,20%	58,70%	96,90%	91,40%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
<i>GK</i>	88,90%	70,40%	100%	99,90%	Glycerol kinase deficiency, 307030
<i>GLB1</i>	99,90%	97,40%	100%	100%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
<i>GLDC</i>	89,90%	82,00%	100%	99,90%	Glycine encephalopathy, 605899
<i>GLI2</i>	99,10%	97,40%	100%	99,80%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
<i>GLI3</i>	100%	99,50%	100%	100%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
<i>GLIS3</i>	100%	99,60%	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
<i>GLUD1</i>	94,20%	82,90%	100%	100%	Hyperinsulinism-hyperammonemia syndrome, 606762
<i>GLYCK</i>	98,80%	97,30%	100%	100%	D-glyceric aciduria, 220120
<i>GM2A</i>	100%	100%	100%	100%	GM2-gangliosidosis, AB variant, 272750
<i>GMPPA</i>	100%	100%	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
<i>GMPPB</i>	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
<i>GNAO1</i>	93,80%	93,80%	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
<i>GNAS</i>	100%	99,90%	100%	99,90%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism 1c, 612462 Pseudohypoparathyroidism 1b, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism 1a, 103580
<i>GNB1</i>	100%	100%	100%	100%	Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
<i>GNB5</i>	100%	98,80%	100%	100%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
<i>GNPAT</i>	99,70%	97,30%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765

<i>GNPTAB</i>	100%	99,90%	100%	100%	Mucopolipidosis II alpha/beta, 252500 Mucopolipidosis III alpha/beta, 252600
<i>GNPTG</i>	99,10%	94,30%	100%	99,90%	Mucopolipidosis III gamma, 252605
<i>GNS</i>	98,40%	94,80%	100%	100%	Mucopolysaccharidosis type IIID, 252940
<i>GPAA1</i>	98,90%	95,90%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
<i>GPC3</i>	99,10%	94,70%	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
<i>GPC4</i>	100%	99,60%	100%	100%	Keipert syndrome, 301026
<i>GPHN</i>	100%	99,50%	100%	100%	Molybdenum cofactor deficiency C, 615501
<i>GPSM2</i>	99,90%	99,20%	100%	100%	Chudley-McCullough syndrome, 604213
<i>GPT2</i>	99,20%	93,60%	100%	99,80%	Mental retardation, autosomal recessive 49, 616281
<i>GRIA3</i>	99,70%	96,10%	100%	99,60%	Mental retardation, X-linked 94, 300699
<i>GRIA4</i>	99,80%	99,00%	100%	100%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
<i>GRID2</i>	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive 18, 616204
<i>GRIK2</i>	96,20%	95,40%	96,30%	96,30%	Mental retardation, autosomal recessive, 6, 611092
<i>GRIN1</i>	100%	100%	100%	100%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
<i>GRIN2A</i>	100%	100%	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
<i>GRIN2B</i>	99,80%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
<i>GRIN2D</i>	79,80%	65,40%	93,90%	88,70%	Epileptic encephalopathy, early infantile, 46, 617162
<i>GRIP1</i>	100%	99,70%	100%	100%	Fraser syndrome 3, 617667
<i>GRM1</i>	100%	99,70%	100%	100%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
<i>GRN</i>	100%	100%	100%	100%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
<i>GSE1</i>	99,80%	97,70%	100%	100%	No OMIM disease ID
<i>GSS</i>	100%	99,90%	100%	100%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
<i>GTF2H5</i>	100%	99,60%	100%	100%	Trichothiodystrophy 3, photosensitive, 616395
<i>GTPBP2</i>	100%	99,30%	100%	99,90%	Jaberi-Elahi syndrome, 617988
<i>GTPBP3</i>	100%	99,80%	100%	100%	Combined oxidative phosphorylation deficiency 23, 616198
<i>GUSB</i>	92,90%	91,70%	100%	100%	Mucopolysaccharidosis VII, 253220
<i>HACE1</i>	100%	99,30%	100%	100%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756

<i>HADH</i>	99,00%	97,50%	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
<i>HADHA</i>	97,10%	91,30%	100%	100%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
<i>HAX1</i>	100%	100%	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
<i>HCCS</i>	99,80%	97,60%	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
<i>HCFC1</i>	98,30%	93,60%	100%	100%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type ), 309541
<i>HCN1</i>	100%	99,70%	100%	100%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
<i>HDAC4</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>HDAC6</i>	99,50%	97,40%	100%	100%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
<i>HDAC8</i>	100%	99,80%	100%	100%	Cornelia de Lange syndrome 5, 300882
<i>HECW2</i>	100%	99,10%	100%	100%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
<i>HEPACAM</i>	86,00%	78,90%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
<i>HERC1</i>	100%	100%	100%	100%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
<i>HERC2</i>	79,90%	77,20%	100%	100%	Mental retardation, autosomal recessive 38, 615516
<i>HESX1</i>	99,70%	97,30%	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
<i>HEXA</i>	93,80%	93,30%	100%	100%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
<i>HEXB</i>	99,60%	96,90%	100%	99,90%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
<i>HGSNAT</i>	86,40%	86,30%	91,20%	89,30%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
<i>HIBCH</i>	98,20%	88,50%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
<i>HIST1H1E</i>	100%	100%	100%	100%	Rahman syndrome, 617537
<i>HIST1H4C</i>	100%	100%	100%	100%	No OMIM disease ID
<i>HIVEP2</i>	100%	100%	100%	100%	Mental retardation, autosomal dominant 43, 616977
<i>HLCS</i>	100%	100%	100%	100%	Holocarboxylase synthetase deficiency, 253270
<i>HMGCL</i>	100%	99,80%	100%	100%	HMG-CoA lyase deficiency, 246450
<i>HNMT</i>	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 51, 616739
<i>HNRNPH2</i>	100%	100%	100%	100%	Mental retardation, X-linked, syndromic, Bain type, 300986
<i>HNRNPK</i>	91,50%	82,80%	100%	100%	Au-Kline syndrome, 616580

<i>HNRNPU</i>	99,90%	98,90%	100%	100%	Epileptic encephalopathy, early infantile, 54, 617391
<i>HOXA1</i>	100%	100%	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
<i>HPD</i>	100%	100%	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
<i>HPRT1</i>	99,30%	91,80%	100%	99,30%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
<i>HRAS</i>	100%	100%	100%	100%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
<i>HSD17B10</i>	100%	99,10%	100%	100%	HSD10 mitochondrial disease, 300438
<i>HSD17B4</i>	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
<i>HSPA9</i>	88,50%	84,50%	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
<i>HSPD1</i>	98,80%	93,70%	100%	100%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
<i>HTRA2</i>	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VIII, 617248
<i>HUWE1</i>	99,20%	95,80%	100%	100%	Mental retardation, X-linked syndromic, Turner type, 309590
<i>HYLS1</i>	100%	100%	100%	100%	Hydrolethalus syndrome, 236680
<i>IARS</i>	100%	99,60%	100%	100%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
<i>IARS2</i>	100%	99,90%	100%	100%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
<i>IDS</i>	99,90%	98,00%	100%	100%	Mucopolysaccharidosis II, 309900
<i>IDUA</i>	93,70%	86,80%	100%	100%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
<i>IER3IP1</i>	91,90%	82,60%	100%	100%	Microcephaly, epilepsy, and diabetes syndrome, 614231
<i>IFIH1</i>	99,70%	98,40%	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
<i>IFT172</i>	99,90%	99,10%	100%	100%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
<i>IFT81</i>	93,50%	90,10%	95,00%	94,90%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
<i>IGBP1</i>	99,50%	96,20%	100%	100%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472

<i>IGF1</i>	100%	99,90%	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
<i>IGF1R</i>	100%	99,90%	100%	100%	Insulin-like growth factor I, resistance to, 270450
<i>IKBKG</i>	84,10%	77,20%	100%	100%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
<i>IL1RAPL1</i>	99,80%	98,60%	100%	100%	Mental retardation, X-linked 21/34, 300143
<i>IMPA1</i>	97,00%	87,00%	100%	100%	Mental retardation, autosomal recessive 59, 617323
<i>INPP5E</i>	97,10%	92,70%	100%	100%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
<i>INPP5K</i>	100%	100%	100%	100%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
<i>MR</i> <i>PS22</i>	99,80%	99,10%	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
<i>INTS1</i>	99,80%	98,50%	100%	100%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
<i>IQSEC1</i>	99,80%	96,70%	100%	99,90%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
<i>IQSEC2</i>	96,80%	88,60%	99,40%	98,40%	Mental retardation, X-linked 1/78, 309530
<i>IRF2BPL</i>	99,50%	95,00%	99,90%	99,20%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
<i>ISCA2</i>	100%	98,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 4, 616370
<i>ISPD</i>	98,50%	94,80%	100%	99,40%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
<i>ITGA7</i>	99,60%	98,00%	100%	100%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
<i>ITPA</i>	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 35, 616647
<i>ITPR1</i>	100%	99,90%	100%	100%	Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
<i>IVD</i>	100%	100%	100%	100%	Isovaleric acidemia, 243500
<i>JAG1</i>	97,70%	96,80%	100%	100%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
<i>JAM3</i>	100%	99,90%	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
<i>JMJD1C</i>	99,90%	99,20%	100%	100%	No OMIM disease ID
<i>KANK1</i>	100%	100%	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
<i>KANSL1</i>	99,90%	99,20%	100%	100%	Koolen-De Vries syndrome, 610443
<i>KAT6A</i>	100%	99,80%	100%	100%	Arboleda-Tham syndrome, 616268
<i>KAT6B</i>	99,90%	99,00%	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170



<i>KATNB1</i>	100%	99,90%	100%	100%	Lissencephaly 6, with microcephaly, 616212
<i>KCNA2</i>	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
<i>KCNA4</i>	100%	100%	100%	100%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
<i>KCNB1</i>	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 26, 616056
<i>KCNC1</i>	100%	100%	100%	100%	Epilepsy, progressive myoclonic 7, 616187
<i>KCNC3</i>	81,10%	69,40%	94,70%	89,00%	Spinocerebellar ataxia 13, 605259
<i>KCNH1</i>	98,70%	98,70%	98,70%	98,70%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
<i>KCNJ10</i>	89,30%	89,00%	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
<i>KCNJ11</i>	100%	100%	100%	100%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
<i>KCNJ6</i>	100%	100%	100%	100%	Keppen-Lubinsky syndrome, 614098
<i>KCNK4</i>	99,10%	97,40%	100%	100%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
<i>KCNK9</i>	100%	100%	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
<i>KCNMA1</i>	94,40%	93,60%	100%	100%	Liang-Wang syndrome, 618729 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
<i>KCNQ2</i>	91,30%	89,80%	100%	100%	Epileptic encephalopathy, early infantile, 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
<i>KCNQ3</i>	100%	99,40%	99,80%	99,10%	Seizures, benign neonatal, 2, 121201
<i>KCNQ5</i>	97,80%	95,50%	100%	100%	Mental retardation, autosomal dominant 46, 617601
<i>KCNT1</i>	96,00%	95,20%	98,60%	97,30%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
<i>KCTD7</i>	95,00%	95,00%	100%	100%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
<i>KDM1A</i>	98,20%	95,20%	100%	100%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
<i>KDM3B</i>	97,50%	96,30%	100%	100%	Diets-Jongmans syndrome, 618846
<i>KDM5B</i>	99,50%	97,90%	97,70%	97,70%	Mental retardation, autosomal recessive 65, 618109
<i>KDM5C</i>	99,80%	97,90%	100%	100%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
<i>KDM6A</i>	96,10%	88,70%	100%	99,90%	Kabuki syndrome 2, 300867
<i>KDM6B</i>	98,80%	97,90%	100%	100%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
<i>KIAA0586</i>	97,30%	93,10%	95,80%	95,80%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
<i>KIAA1109</i>	99,80%	99,20%	100%	100%	Alkuraya-Kucinskas syndrome, 617822

<i>KIDINS220</i>	100%	100%	100%	100%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
<i>KIF11</i>	97,60%	94,80%	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
<i>KIF14</i>	99,60%	97,70%	100%	100%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
<i>KIF1A</i>	99,40%	97,10%	100%	100%	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
<i>KIF1BP</i>	96,10%	96,10%	96,10%	96,10%	Goldberg-Shprintzen megacolon syndrome, 609460
<i>KIF2A</i>	99,60%	95,60%	100%	100%	Cortical dysplasia, complex, with other brain malformations 3, 615411
<i>KIF4A</i>	99,40%	95,70%	100%	100%	?Mental retardation, X-linked 100, 300923
<i>KIF5C</i>	99,90%	98,80%	99,80%	99,80%	Cortical dysplasia, complex, with other brain malformations 2, 615282
<i>KIF7</i>	93,60%	90,60%	99,10%	97,80%	?Hydroletharus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
<i>KIRREL3</i>	99,80%	98,90%	100%	100%	No OMIM disease ID
<i>KLF7</i>	100%	99,70%	100%	100%	No OMIM disease ID
<i>KLHL15</i>	100%	99,70%	100%	100%	Mental retardation, X-linked 103, 300982
<i>KMT2A</i>	100%	99,90%	99,90%	99,40%	Wiedemann-Steiner syndrome, 605130
<i>KMT2B</i>	95,80%	94,00%	98,70%	97,90%	Dystonia 28, childhood-onset, 617284
<i>KMT2C</i>	92,20%	91,00%	100%	100%	Kleefstra syndrome 2, 617768
<i>KMT2D</i>	100%	99,40%	100%	100%	Kabuki syndrome 1, 147920
<i>KMT2E</i>	99,80%	98,50%	100%	100%	O'Donnell-Luria-Rodan syndrome, 618512
<i>KMT5B</i>	99,90%	99,10%	100%	100%	Mental retardation, autosomal dominant 51, 617788
<i>KNL1</i>	99,20%	98,10%	98,90%	98,80%	Microcephaly 4, primary, autosomal recessive, 604321
<i>KPTN</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 41, 615637
<i>KRAS</i>	99,50%	96,90%	100%	100%	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215

					Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
<i>L1CAM</i>	99,90%	99,10%	100%	100%	MASA syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000
<i>L2HGDH</i>	99,00%	97,20%	100%	100%	L-2-hydroxyglutaric aciduria, 236792
<i>LAMA1</i>	100%	99,70%	100%	100%	Poretti-Boltshauser syndrome, 615960
<i>LAMA2</i>	100%	99,60%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
<i>LAMB1</i>	100%	99,90%	100%	100%	Lissencephaly 5, 615191
<i>LAMC3</i>	98,60%	97,10%	100%	99,60%	Cortical malformations, occipital, 614115
<i>LAMP2</i>	99,20%	95,60%	100%	100%	Danon disease, 300257
<i>LARGE1</i>	100%	99,60%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
<i>LARP7</i>	88,50%	78,40%	100%	100%	Alazami syndrome, 615071
<i>LAS1L</i>	99,70%	97,30%	100%	100%	Wilson-Turner syndrome, 309585
<i>LIAS</i>	100%	99,10%	100%	100%	Hyperglycinemia, lactic acidosis, and seizures, 614462
<i>LIG4</i>	100%	99,90%	100%	100%	LIG4 syndrome, 606593
<i>LINGO1</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 64, 618103
<i>LINS1</i>	100%	99,10%	100%	100%	Mental retardation, autosomal recessive 27, 614340
<i>LMAN2L</i>	100%	99,70%	100%	100%	?Mental retardation, autosomal recessive, 52, 616887
<i>LONP1</i>	100%	99,80%	100%	100%	CODAS syndrome, 600373
<i>LRP2</i>	100%	99,90%	100%	100%	Donnai-Barrow syndrome, 222448
<i>LRPPRC</i>	99,90%	99,10%	100%	100%	Leigh syndrome, French-Canadian type, 220111
<i>LYST</i>	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
<i>LZTFL1</i>	99,90%	99,20%	100%	100%	Bardet-Biedl syndrome 17, 615994
<i>LZTR1</i>	100%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
<i>MAB21L1</i>	100%	100%	100%	100%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
<i>MAB21L2</i>	100%	100%	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
<i>MACF1</i>	100%	99,60%	100%	100%	Lissencephaly 9 with complex brainstem malformation, 618325
<i>MAF</i>	83,50%	78,00%	88,60%	82,20%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
<i>MAG</i>	100%	100%	100%	100%	Spastic paraplegia 75, autosomal recessive, 616680

<i>MAGEL2</i>	93,00%	87,20%	100%	100%	Schaaf-Yang syndrome, 615547
<i>MAN1B1</i>	100%	99,70%	100%	99,90%	Mental retardation, autosomal recessive 15, 614202
<i>MAN2B1</i>	99,80%	97,90%	100%	100%	Mannosidosis, alpha-, types I and II, 248500
<i>MANBA</i>	99,80%	98,40%	100%	100%	Mannosidosis, beta, 248510
<i>MAOA</i>	100%	99,70%	99,80%	98,50%	Brunner syndrome, 300615
<i>MAP1B</i>	99,30%	97,70%	100%	100%	No OMIM disease ID
<i>MAP2K1</i>	99,80%	97,10%	100%	100%	Cardiofaciocutaneous syndrome 3, 615279
<i>MAP2K2</i>	98,50%	95,10%	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
<i>MAPK8IP3</i>	100%	99,60%	100%	100%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
<i>MAPRE2</i>	100%	99,30%	100%	100%	Symmetric circumferential skin creases, congenital, 2, 616734
<i>MASP1</i>	100%	99,90%	100%	100%	3MC syndrome 1, 257920
<i>MAST1</i>	100%	99,50%	100%	100%	Mega-corporum-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
<i>MAT1A</i>	99,70%	97,70%	100%	100%	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
<i>MBD5</i>	99,90%	99,90%	100%	100%	Mental retardation, autosomal dominant 1, 156200
<i>MBOAT7</i>	100%	99,50%	100%	100%	Mental retardation, autosomal recessive 57, 617188
<i>MBTPS2</i>	100%	99,00%	100%	100%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
<i>MCCC1</i>	100%	99,80%	100%	100%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
<i>MCCC2</i>	99,90%	98,40%	100%	100%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
<i>MCOLN1</i>	99,80%	98,40%	100%	100%	Mucopolipidosis IV, 252650
<i>MCPH1</i>	100%	99,40%	100%	100%	Microcephaly 1, primary, autosomal recessive, 251200
<i>MDH2</i>	98,00%	97,90%	100%	100%	Epileptic encephalopathy, early infantile, 51, 617339
<i>MECP2</i>	100%	98,70%	100%	99,90%	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
<i>MECR</i>	100%	98,90%	100%	100%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
<i>MED12</i>	99,80%	96,70%	100%	100%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
<i>MED13</i>	100%	99,90%	100%	100%	Intellectual developmental disorder 61, 618009

<i>MED13L</i>	100%	99,80%	100%	100%	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
<i>MED17</i>	96,30%	93,50%	100%	100%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
<i>MED23</i>	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 18, 614249
<i>MED25</i>	100%	99,80%	100%	100%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
<i>MEF2C</i>	99,90%	96,00%	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
<i>MEGF8</i>	99,90%	99,00%	100%	100%	Carpenter syndrome 2, 614976
<i>MEIS2</i>	100%	100%	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
<i>METTL23</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 44, 615942
<i>METTL5</i>	99,30%	98,50%	99,90%	98,30%	Intellectual developmental disorder, autosomal recessive 72, 618665
<i>MFF</i>	94,30%	89,90%	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
<i>MFSD2A</i>	99,70%	98,50%	100%	100%	Microcephaly 15, primary, autosomal recessive, 616486
<i>MFSD8</i>	100%	99,70%	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
<i>MGAT2</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
<i>MGP</i>	98,70%	95,10%	100%	100%	Keutel syndrome, 245150
<i>MICU1</i>	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
<i>MID1</i>	99,80%	98,70%	100%	100%	Opitz GBBB syndrome, type I, 300000
<i>MID2</i>	99,80%	98,70%	100%	100%	?Mental retardation, X-linked 101, 300928
<i>MKKS</i>	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
<i>MKS1</i>	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
<i>MLC1</i>	100%	99,00%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
<i>MLYCD</i>	96,00%	90,40%	100%	98,90%	Malonyl-CoA decarboxylase deficiency, 248360
<i>MMAA</i>	100%	100%	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
<i>MMAB</i>	100%	99,60%	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
<i>MMACHC</i>	100%	100%	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
<i>MMADHC</i>	94,40%	83,50%	89,70%	89,70%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
<i>MN1</i>	100%	99,30%	100%	100%	Meningioma, 607174 CEBALID syndrome, 618774
<i>MOCS1</i>	99,20%	95,40%	100%	100%	Molybdenum cofactor deficiency A, 252150

<i>MOCS2</i>	99,60%	99,50%	100%	100%	Molybdenum cofactor deficiency B, 252160
<i>MOGS</i>	100%	99,90%	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
<i>MPDU1</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type If, 609180
<i>MPDZ</i>	99,80%	98,80%	100%	100%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
<i>MPLKIP</i>	100%	99,40%	100%	100%	Trichothiodystrophy 4, nonphotosensitive, 234050
<i>MSL3</i>	97,00%	88,30%	100%	100%	Basilicata-Akhtar syndrome, 301032
<i>MSMO1</i>	96,30%	88,90%	100%	100%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
<i>MTFMT</i>	100%	99,80%	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
<i>MTHFR</i>	97,30%	96,00%	100%	100%	Homocystinuria due to MTHFR deficiency, 236250
<i>MTOR</i>	100%	99,50%	100%	100%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
<i>MTR</i>	100%	100%	100%	100%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
<i>MTRR</i>	100%	99,60%	100%	100%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
<i>MUT</i>	99,80%	98,30%	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
<i>MVK</i>	90,90%	90,50%	90,50%	90,50%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
<i>MYCN</i>	100%	99,90%	99,30%	96,70%	Feingold syndrome 1, 164280
<i>MYH9</i>	100%	99,30%	100%	100%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
<i>MYO5A</i>	99,80%	98,90%	100%	100%	Griscelli syndrome, type 1, 214450
<i>MYT1L</i>	99,80%	99,00%	100%	100%	Mental retardation, autosomal dominant 39, 616521
<i>NAA10</i>	99,70%	98,50%	99,90%	99,90%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
<i>NAA15</i>	95,80%	91,00%	96,80%	96,70%	Mental retardation, autosomal dominant 50, 617787
<i>NACC1</i>	100%	99,80%	100%	100%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
<i>NAGA</i>	100%	100%	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
<i>NAGLU</i>	92,90%	89,90%	99,90%	99,20%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
<i>NALCN</i>	100%	99,50%	99,80%	99,80%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
<i>NANS</i>	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442

<i>NARS2</i>	98,30%	97,40%	100%	100%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
<i>NAXE</i>	100%	99,80%	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
<i>NBEA</i>	92,00%	90,60%	100%	100%	No OMIM disease ID
<i>NBN</i>	99,90%	98,60%	100%	100%	Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260 Leukemia, acute lymphoblastic, 613065
<i>NCAPG2</i>	99,90%	99,20%	100%	100%	Khan-Khan-Katsanis syndrome, 618460
<i>NDE1</i>	100%	100%	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
<i>NDP</i>	100%	99,70%	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
<i>NDST1</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 46, 616116
<i>NDUFA1</i>	99,90%	99,30%	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020
<i>NDUFA11</i>	100%	100%	100%	99,80%	Mitochondrial complex I deficiency, nuclear type 14, 618236
<i>NDUFA12</i>	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
<i>NDUFA2</i>	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
<i>NDUF3</i>	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
<i>NDUF5</i>	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
<i>NDUF8</i>	62,60%	61,10%	100%	99,60%	Mitochondrial complex I deficiency, nuclear type 34, 618776
<i>NDUFS1</i>	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
<i>NDUFS2</i>	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
<i>NDUFS3</i>	90,70%	90,60%	91,90%	90,70%	Mitochondrial complex I deficiency, nuclear type 8, 618230
<i>NDUFS4</i>	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
<i>NDUFS6</i>	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
<i>NDUFS7</i>	100%	99,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 3, 618224
<i>NDUFS8</i>	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 2, 618222
<i>NDUFV1</i>	98,00%	96,10%	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
<i>NDUFV2</i>	86,90%	76,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
<i>NEDD4L</i>	72,00%	71,50%	100%	100%	Periventricular nodular heterotopia 7, 617201
<i>NEU1</i>	99,70%	97,70%	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
<i>NEXMIF</i>	100%	99,50%	100%	100%	Mental retardation, X-linked 98, 300912
<i>NF1</i>	92,60%	90,20%	100%	100%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321

					Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210
<i>NFIA</i>	100%	99,60%	100%	100%	Brain malformations with or without urinary tract defects, 613735
<i>NFIB</i>	97,40%	96,50%	100%	100%	Macrocephaly, acquired, with impaired intellectual development, 618286
<i>NFIX</i>	100%	99,50%	99,60%	98,70%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
<i>NFU1</i>	98,80%	90,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
<i>NGLY1</i>	100%	99,80%	100%	100%	Congenital disorder of deglycosylation, 615273
<i>NHS</i>	95,40%	93,90%	100%	99,80%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
<i>NIPBL</i>	98,90%	97,00%	100%	100%	Cornelia de Lange syndrome 1, 122470
<i>NKAP</i>	99,30%	95,20%	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
<i>NKX2-1</i>	98,60%	85,60%	100%	100%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
<i>NLGN3</i>	99,90%	99,00%	100%	100%	No OMIM disease ID
<i>NLGN4X</i>	99,90%	98,90%	100%	99,90%	Mental retardation, X-linked, 300495
<i>NONO</i>	100%	98,40%	100%	100%	Mental retardation, X-linked, syndromic 34, 300967
<i>NOVA2</i>	99,00%	94,60%	96,80%	93,30%	No OMIM disease ID
<i>NPC1</i>	99,60%	98,70%	100%	100%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
<i>NPC2</i>	100%	99,60%	100%	100%	Niemann-pick disease, type C2, 607625
<i>NPHP1</i>	100%	99,00%	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
<i>NR2F1</i>	100%	100%	99,10%	95,10%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
<i>NR4A2</i>	100%	100%	100%	100%	No OMIM disease ID
<i>NRAS</i>	100%	100%	100%	100%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaicism, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
<i>NRXN1</i>	97,40%	96,90%	100%	99,80%	Pitt-Hopkins-like syndrome 2, 614325
<i>NSD1</i>	100%	99,90%	100%	100%	Sotos syndrome 1, 117550
<i>NSD2</i>	99,90%	99,20%	100%	100%	No OMIM disease ID



<i>NSDHL</i>	100%	98,70%	100%	100%	CHILD syndrome, 308050 CK syndrome, 300831
<i>NSUN2</i>	96,00%	95,10%	100%	100%	Mental retardation, autosomal recessive 5, 611091
<i>NT5C2</i>	98,00%	96,50%	100%	100%	Spastic paraplegia 45, autosomal recessive, 613162
<i>NTNG2</i>	98,50%	96,70%	99,90%	99,00%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
<i>NTRK1</i>	99,80%	98,20%	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
<i>NTRK2</i>	100%	99,90%	100%	100%	Obesity, hyperphagia, and developmental delay, 613886 Epileptic encephalopathy, early infantile, 58, 617830
<i>NUBPL</i>	99,70%	98,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 21, 618242
<i>NUP62</i>	100%	100%	100%	100%	Striatonigral degeneration, infantile, 271930
<i>NUS1</i>	60,00%	44,50%	100%	100%	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
<i>OAT</i>	85,20%	76,30%	100%	100%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
<i>OCLN</i>	100%	100%	100%	100%	Pseudo-TORCH syndrome 1, 251290
<i>OCRL</i>	99,90%	98,60%	100%	99,90%	Low syndrome, 309000 Dent disease 2, 300555
<i>ODC1</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>OFD1</i>	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
<i>OGT</i>	99,90%	99,00%	100%	100%	Mental retardation, X-linked 106, 300997
<i>OPHN1</i>	99,50%	97,60%	99,90%	98,80%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
<i>ORC1</i>	100%	99,40%	100%	100%	Meier-Gorlin syndrome 1, 224690
<i>OSGEP</i>	100%	99,40%	100%	100%	Galloway-Mowat syndrome 3, 617729
<i>OTC</i>	100%	100%	100%	100%	Ornithine transcarbamylase deficiency, 311250
<i>OTUD6B</i>	99,90%	98,80%	100%	100%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
<i>OTX2</i>	100%	99,70%	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
<i>OXR1</i>	99,40%	97,00%	100%	100%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
<i>P4HTM</i>	99,00%	97,40%	100%	99,40%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
<i>PACS1</i>	98,80%	96,90%	100%	100%	Schuurs-Hoeijmakers syndrome, 615009
<i>PACS2</i>	99,30%	96,20%	100%	99,80%	Epileptic encephalopathy, early infantile, 66, 618067
<i>PAFAH1B1</i>	94,10%	87,10%	100%	100%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432

<i>PAH</i>	100%	100%	100%	100%	Phenylketonuria, 261600
<i>PAK1</i>	100%	99,60%	100%	100%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
<i>PAK3</i>	99,30%	95,90%	100%	99,80%	Mental retardation, X-linked 30/47, 300558
<i>PANK2</i>	100%	99,30%	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
<i>PANX1</i>	100%	100%	100%	100%	Oocyte maturation defect 7, 618550
<i>PARN</i>	100%	99,90%	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
<i>PAX1</i>	92,40%	87,90%	100%	99,60%	Otofaciocervical syndrome 2, 615560
<i>PAX6</i>	100%	100%	100%	100%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Cataract with late-onset corneal dystrophy, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229
<i>PAX7</i>	100%	100%	100%	100%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
<i>PAX8</i>	100%	99,80%	100%	100%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
<i>PBX1</i>	100%	99,40%	100%	100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
<i>PC</i>	99,80%	97,30%	100%	100%	Pyruvate carboxylase deficiency, 266150
<i>PCCA</i>	99,50%	96,70%	100%	100%	Propionicacidemia, 606054
<i>PCCB</i>	97,90%	96,00%	98,70%	96,20%	Propionicacidemia, 606054
<i>PCDH12</i>	100%	100%	100%	100%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
<i>PCDH19</i>	100%	98,90%	100%	100%	Epileptic encephalopathy, early infantile, 9, 300088
<i>PCGF2</i>	100%	99,50%	100%	100%	Turnpenny-Fry syndrome, 618371
<i>PCLO</i>	99,70%	98,70%	100%	100%	?Pontocerebellar hypoplasia, type 3, 608027
<i>PCNT</i>	99,60%	97,10%	100%	100%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
<i>PDE4D</i>	95,70%	93,50%	100%	99,80%	Acrodysostosis 2, with or without hormone resistance, 614613
<i>PDHA1</i>	99,40%	97,10%	100%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
<i>PDHX</i>	99,90%	99,40%	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
<i>PDP1</i>	100%	100%	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
<i>PDSS1</i>	94,70%	87,60%	97,30%	96,60%	Coenzyme Q10 deficiency, primary, 2, 614651
<i>PDSS2</i>	99,80%	97,10%	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
<i>PEPD</i>	100%	98,80%	100%	100%	Prolidase deficiency, 170100

<i>PET100</i>	100%	99,60%	100%	100%	Mitochondrial complex IV deficiency, 220110
<i>PEX1</i>	99,90%	99,40%	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
<i>PEX10</i>	96,80%	89,70%	100%	99,90%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
<i>PEX11B</i>	100%	99,60%	100%	100%	?Peroxisome biogenesis disorder 14B, 614920
<i>PEX12</i>	100%	100%	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
<i>PEX13</i>	100%	100%	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
<i>PEX16</i>	97,90%	94,20%	100%	100%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
<i>PEX19</i>	99,90%	98,50%	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
<i>PEX2</i>	100%	100%	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
<i>PEX26</i>	100%	100%	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
<i>PEX3</i>	100%	99,30%	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
<i>PEX5</i>	99,90%	99,00%	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
<i>PEX6</i>	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
<i>PEX7</i>	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
<i>PGAP1</i>	99,00%	94,40%	100%	100%	Mental retardation, autosomal recessive 42, 615802
<i>PGAP2</i>	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
<i>PGAP3</i>	63,50%	59,60%	100%	100%	Hyperphosphatasia with mental retardation syndrome 4, 615716
<i>PGK1</i>	92,80%	79,30%	100%	100%	Phosphoglycerate kinase 1 deficiency, 300653
<i>PGM3</i>	100%	99,80%	100%	100%	Immunodeficiency 23, 615816
<i>PHACTR1</i>	100%	99,70%	100%	100%	Epileptic encephalopathy, early infantile, 70, 618298
<i>PHF21A</i>	100%	99,90%	100%	100%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
<i>PHF6</i>	97,80%	88,30%	99,90%	98,90%	Borjeson-Forssman-Lehmann syndrome, 301900
<i>PHF8</i>	99,70%	96,80%	100%	100%	Mental retardation syndrome, X-linked, Siderius type, 300263

<i>PHGDH</i>	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
<i>PHIP</i>	98,60%	96,10%	100%	99,70%	Chung-Jansen syndrome, 617991
<i>PI4KA</i>	92,60%	88,80%	99,90%	99,90%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogyposis, 616531
<i>TAF1</i>	99,80%	97,70%	100%	100%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
<i>TAF13</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 60, 617432
<i>TAF2</i>	99,90%	98,60%	100%	100%	Mental retardation, autosomal recessive 40, 615599
<i>TAF6</i>	99,80%	98,90%	100%	100%	Alazami-Yuan syndrome, 617126
<i>TANC2</i>	100%	99,50%	100%	100%	No OMIM disease ID
<i>TANGO2</i>	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
<i>TAOK1</i>	99,50%	97,90%	100%	100%	No OMIM disease ID
<i>TASP1</i>	99,70%	98,80%	100%	100%	No OMIM disease ID
<i>TAT</i>	100%	100%	100%	100%	Tyrosinemia, type II, 276600
<i>TBC1D20</i>	94,20%	94,20%	100%	99,90%	Warburg micro syndrome 4, 615663
<i>TBC1D23</i>	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 11, 617695
<i>PIGA</i>	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
<i>PIGB</i>	99,90%	97,80%	100%	100%	Epileptic encephalopathy, early infantile, 80, 618580
<i>PIGC</i>	99,20%	90,90%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
<i>PIGG</i>	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 53, 616917
<i>PIGL</i>	100%	100%	100%	100%	CHIME syndrome, 280000
<i>PIGN</i>	93,80%	91,50%	98,80%	98,80%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
<i>PIGO</i>	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
<i>PIGP</i>	95,80%	87,30%	100%	100%	Epileptic encephalopathy, early infantile, 55, 617599
<i>PIGT</i>	98,10%	98,10%	100%	100%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
<i>PIGU</i>	100%	99,10%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 21, 618590
<i>PIGV</i>	100%	100%	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
<i>PIGW</i>	100%	99,80%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
<i>PIGY</i>	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 6, 616809
<i>PIK3CA</i>	100%	99,80%	100%	100%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550

					Breast cancer, somatic, 114480 Macroductyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
<i>PIK3R2</i>	90,70%	89,60%	99,30%	96,10%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
<i>PLA2G6</i>	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
<i>PLAA</i>	100%	99,20%	100%	100%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
<i>PLCB1</i>	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
<i>PLK4</i>	99,90%	98,20%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
<i>PLP1</i>	100%	99,20%	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
<i>PLPBP</i>	98,20%	90,10%	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
<i>PLXND1</i>	98,90%	96,20%	99,70%	99,40%	No OMIM disease ID
<i>PMM2</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
<i>PMPCA</i>	97,70%	94,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 2, 213200
<i>PMPCB</i>	100%	99,70%	100%	100%	Multiple mitochondrial dysfunctions syndrome 6, 617954
<i>PNKP</i>	100%	100%	100%	100%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
<i>PNP</i>	99,80%	98,90%	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
<i>PNPLA6</i>	100%	99,70%	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
<i>POGZ</i>	99,40%	99,00%	100%	100%	White-Sutton syndrome, 616364
<i>POLA1</i>	99,30%	95,40%	100%	99,90%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
<i>POLG</i>	100%	99,30%	100%	100%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450

<i>POLR1C</i>	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
<i>POLR2A</i>	100%	100%	100%	100%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
<i>POLR3A</i>	100%	99,70%	100%	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
<i>POLR3B</i>	99,90%	98,60%	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
<i>POMGNT1</i>	100%	99,90%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
<i>POMGNT2</i>	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
<i>POMK</i>	100%	100%	100%	100%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
<i>POMT1</i>	99,30%	97,50%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
<i>POMT2</i>	99,40%	96,40%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
<i>PORCN</i>	100%	99,10%	100%	100%	Focal dermal hypoplasia, 305600
<i>POU1F1</i>	100%	99,20%	100%	100%	Pituitary hormone deficiency, combined, 1, 613038
<i>POU3F3</i>	73,20%	59,60%	94,90%	83,80%	Snijders Blok-Fisher syndrome, 618604
<i>PPM1D</i>	100%	99,90%	100%	100%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
<i>PPP1CB</i>	99,90%	99,30%	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
<i>PPP1R12A</i>	97,70%	95,30%	100%	100%	Genitourinary and/or/brain malformation syndrome, 618820
<i>PPP1R15B</i>	100%	99,60%	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
<i>PPP1R21</i>	99,60%	96,00%	100%	100%	No OMIM disease ID
<i>PPP2CA</i>	100%	100%	100%	100%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
<i>PPP2R1A</i>	91,60%	91,50%	93,60%	93,60%	Mental retardation, autosomal dominant 36, 616362
<i>PPP2R5B</i>	100%	100%	100%	100%	No OMIM disease ID
<i>PPP2R5C</i>	97,70%	93,10%	100%	100%	No OMIM disease ID
<i>PPP2R5D</i>	100%	100%	100%	100%	Mental retardation, autosomal dominant 35, 616355
<i>PPP3CA</i>	99,80%	98,40%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 1, 617711 Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265

<i>PPT1</i>	90,30%	90,30%	82,50%	82,50%	Ceroid lipofuscinosis, neuronal, 1, 256730
<i>PQBP1</i>	100%	100%	100%	100%	Renpenning syndrome, 309500
<i>PRKAR1A</i>	99,30%	93,50%	100%	100%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
<i>PRMT7</i>	100%	99,90%	100%	100%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
<i>PRODH</i>	85,00%	80,60%	100%	100%	Hyperprolinemia, type I, 239500
<i>PRPS1</i>	100%	100%	100%	100%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
<i>PRR12</i>	98,70%	97,20%	100%	100%	No OMIM disease ID
<i>PRSS12</i>	100%	99,90%	100%	100%	Mental retardation, autosomal recessive 1, 249500
<i>PRUNE1</i>	100%	99,90%	100%	100%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
<i>PSAP</i>	100%	100%	100%	100%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
<i>PSAT1</i>	95,30%	81,60%	100%	100%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
<i>PSMD12</i>	98,60%	92,90%	100%	100%	Stankiewicz-Isidor syndrome, 617516
<i>PSPH</i>	100%	100%	100%	100%	Phosphoserine phosphatase deficiency, 614023
<i>PTCH1</i>	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoencephaly 7, 610828
<i>PTCHD1</i>	100%	99,90%	100%	100%	No OMIM disease ID
<i>PTDSS1</i>	100%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
<i>PTEN</i>	99,50%	97,00%	100%	100%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
<i>PTF1A</i>	95,80%	85,60%	98,60%	93,30%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
<i>PTPN11</i>	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250

					Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
<i>PTRH2</i>	100%	100%	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
<i>PTRHD1</i>	100%	100%	100%	100%	No OMIM disease ID
<i>PTS</i>	99,90%	99,10%	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
<i>PUF60</i>	100%	99,30%	100%	100%	Verheij syndrome, 615583
<i>PUM1</i>	100%	99,90%	100%	100%	Spinocerebellar ataxia 47, 617931
<i>PURA</i>	99,00%	95,20%	100%	99,80%	Mental retardation, autosomal dominant 31, 616158
<i>PUS1</i>	100%	99,50%	99,60%	97,20%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
<i>PUS3</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 55, 617051
<i>PUS7</i>	100%	99,80%	100%	100%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
<i>PYCR1</i>	99,90%	97,70%	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
<i>PYCR2</i>	100%	99,10%	100%	100%	Leukodystrophy, hypomyelinating, 10, 616420
<i>QARS</i>	100%	100%	100%	100%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
<i>QDPR</i>	100%	99,70%	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
<i>QRICH1</i>	100%	99,90%	100%	100%	Ververi-Brady syndrome, 617982
<i>RAB11B</i>	100%	100%	100%	100%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
<i>RAB18</i>	99,50%	97,40%	100%	100%	Warburg micro syndrome 3, 614222
<i>RAB23</i>	100%	99,50%	100%	100%	Carpenter syndrome, 201000
<i>RAB27A</i>	100%	100%	100%	100%	Griscelli syndrome, type 2, 607624
<i>RAB39B</i>	100%	100%	100%	100%	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
<i>RAB3GAP1</i>	99,40%	98,90%	99,40%	99,40%	Warburg micro syndrome 1, 600118
<i>RAB3GAP2</i>	99,50%	97,00%	100%	100%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
<i>RAC1</i>	99,90%	96,20%	100%	99,90%	Mental retardation, autosomal dominant 48, 617751
<i>RAC3</i>	97,30%	94,40%	99,70%	98,20%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
<i>RAD21</i>	99,20%	96,60%	100%	100%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
<i>RAF1</i>	100%	100%	100%	100%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
<i>RAI1</i>	100%	100%	100%	100%	Smith-Magenis syndrome, 182290
<i>RALA</i>	94,60%	87,90%	100%	100%	No OMIM disease ID
<i>RALGAPA1</i>	74,50%	63,90%	100%	100%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797



<i>RARB</i>	100%	100%	100%	100%	Microphthalmia, syndromic 12, 615524
<i>RARS</i>	94,20%	91,60%	94,40%	94,30%	Leukodystrophy, hypomyelinating, 9, 616140
<i>RARS2</i>	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
<i>RBBP8</i>	100%	99,70%	100%	100%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
<i>RBFOX1</i>	89,20%	88,80%	99,20%	97,70%	No OMIM disease ID
<i>RBM10</i>	99,50%	97,10%	100%	100%	TARP syndrome, 311900
<i>RBM28</i>	100%	100%	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
<i>RBPJ</i>	98,40%	92,80%	100%	100%	Adams-Oliver syndrome 3, 614814
<i>RCBTB1</i>	99,90%	99,50%	100%	100%	Retinal dystrophy with or without extraocular anomalies, 617175
<i>RECQL4</i>	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
<i>RELN</i>	100%	99,80%	100%	100%	Lissencephaly 2 (Norman-Roberts type), 257320
<i>RERE</i>	96,30%	91,40%	100%	100%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
<i>REV3L</i>	97,60%	97,20%	97,60%	97,60%	No OMIM disease ID
<i>RFT1</i>	99,80%	99,60%	100%	100%	Congenital disorder of glycosylation, type In, 612015
<i>RFX3</i>	100%	100%	100%	100%	No OMIM disease ID
<i>RHEB</i>	88,80%	75,40%	100%	100%	No OMIM disease ID
<i>RHOBTB2</i>	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 64, 618004
<i>RMRP</i>	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
<i>RIT1</i>	100%	100%	100%	100%	Noonan syndrome 8, 615355
<i>RLIM</i>	100%	99,00%	100%	100%	Tonne-Kalscheuer syndrome, 300978
<i>RMND1</i>	100%	98,60%	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922
<i>RNASEH2A</i>	100%	100%	100%	100%	Aicardi-Goutieres syndrome 4, 610333
<i>RNASEH2B</i>	96,00%	92,50%	100%	99,80%	Aicardi-Goutieres syndrome 2, 610181
<i>RNASEH2C</i>	100%	99,50%	100%	100%	Aicardi-Goutieres syndrome 3, 610329
<i>RNASET2</i>	97,40%	93,10%	100%	100%	Leukoencephalopathy, cystic, without megalencephaly, 612951
<i>RNF113A</i>	100%	100%	100%	100%	?Trichothiodystrophy 5, nonphotosensitive, 300953
<i>RNF125</i>	99,90%	99,20%	100%	100%	Tenorio syndrome, 616260
<i>RNF13</i>	95,20%	81,60%	100%	100%	Epileptic encephalopathy, early infantile, 73, 618379
<i>ROGDI</i>	98,40%	95,20%	99,90%	99,10%	Kohlschutter-Tonz syndrome, 226750
<i>ROR2</i>	100%	99,90%	97,00%	97,00%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310

<i>RORA</i>	96,70%	90,20%	100%	100%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
<i>RPGRI1L</i>	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
<i>RPL10</i>	97,40%	89,10%	100%	100%	Mental retardation, X-linked, syndromic, 35, 300998
<i>RPS19</i>	100%	99,60%	100%	100%	Diamond-Blackfan anemia 1, 105650
<i>RPS6KA3</i>	98,40%	94,40%	99,90%	98,80%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
<i>RRM2B</i>	100%	99,70%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
<i>RSPRY1</i>	100%	100%	100%	100%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
<i>RSRC1</i>	99,80%	96,80%	100%	100%	Intellectual developmental disorder, autosomal recessive 70, 618402
<i>RTEL1</i>	99,50%	96,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
<i>RTN4IP1</i>	99,90%	98,70%	100%	100%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
<i>RTTN</i>	98,90%	98,00%	100%	100%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
<i>RUBCN</i>	99,40%	97,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
<i>RUSC2</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 61, 617773
<i>SALL1</i>	99,90%	99,00%	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
<i>SAMD9</i>	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
<i>SAMHD1</i>	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
<i>SARS</i>	100%	99,30%	100%	100%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
<i>SATB2</i>	99,70%	97,40%	100%	100%	Glass syndrome, 612313
<i>SBDS</i>	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
<i>SC5D</i>	100%	99,50%	100%	100%	Lathosterolosis, 607330
<i>SCAMP5</i>	100%	100%	100%	100%	No OMIM disease ID
<i>SCAPER</i>	99,70%	98,20%	100%	100%	Intellectual developmental disorder and retinitis pigmentosa, 618195
<i>SCN1A</i>	99,90%	99,50%	100%	100%	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208
<i>SCN1B</i>	98,00%	96,40%	99,80%	99,30%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377

					Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	99,60%	97,60%	100%	100%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	99,90%	99,30%	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	100%	99,80%	100%	100%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SCO1	97,10%	93,80%	100%	100%	Mitochondrial complex IV deficiency, 220110
SCO2	100%	100%	100%	100%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCYL1	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	100%	99,90%	100%	100%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	85,80%	80,40%	100%	100%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SEMA3E	100%	99,60%	100%	100%	?CHARGE syndrome, 214800
SEPSECS	100%	100%	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,90%	99,50%	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	98,30%	90,50%	98,90%	97,00%	Mental retardation, autosomal dominant 58, 618106
SETBP1	99,90%	98,70%	100%	100%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	100%	99,80%	100%	100%	Epilepsy, early-onset, with or without developmental delay, 618832
SETD1B	98,20%	97,50%	100%	100%	No OMIM disease ID
SETD2	100%	99,90%	100%	100%	Luscan-Lumish syndrome, 616831
SETD5	100%	99,80%	100%	100%	Mental retardation, autosomal dominant 23, 615761
SGPL1	100%	100%	100%	100%	Nephrotic syndrome, type 14, 617575
SGSH	94,40%	94,10%	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK2	100%	99,80%	100%	100%	No OMIM disease ID
SHANK3	92,20%	82,10%	98,00%	94,40%	Phelan-McDermid syndrome, 606232
SHH	100%	99,50%	100%	100%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638

					Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
<i>SHOC2</i>	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
<i>SHROOM4</i>	100%	99,00%	100%	100%	Stocco dos Santos X-linked mental retardation syndrome, 300434
<i>SIK1</i>	98,70%	94,40%	100%	100%	Epileptic encephalopathy, early infantile, 30, 616341
<i>SIL1</i>	99,20%	96,70%	100%	100%	Marinesco-Sjogren syndrome, 248800
<i>SIN3A</i>	100%	99,00%	100%	100%	Witteveen-Kolk syndrome, 613406
<i>SIX3</i>	99,90%	98,60%	100%	100%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
<i>SKI</i>	99,30%	94,90%	100%	99,40%	Shprintzen-Goldberg syndrome, 182212
<i>SLC12A5</i>	83,90%	83,80%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 34, 616645
<i>SLC12A6</i>	100%	100%	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
<i>SLC13A5</i>	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905
<i>SLC16A2</i>	99,20%	93,70%	100%	100%	Allan-Herndon-Dudley syndrome, 300523
<i>SLC17A5</i>	99,60%	97,00%	100%	100%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
<i>SLC19A3</i>	100%	99,80%	98,70%	98,70%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
<i>SLC1A1</i>	99,90%	99,60%	100%	100%	Dicarboxylic aminoaciduria, 222730
<i>SLC1A2</i>	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 41, 617105
<i>SLC1A4</i>	99,00%	95,80%	100%	100%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
<i>SLC25A1</i>	95,80%	88,60%	99,50%	97,80%	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
<i>SLC25A12</i>	99,90%	99,50%	100%	100%	Epileptic encephalopathy, early infantile, 39, 612949
<i>SLC25A15</i>	99,80%	98,10%	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
<i>SLC25A22</i>	98,60%	95,80%	100%	100%	Epileptic encephalopathy, early infantile, 3, 609304
<i>SLC25A24</i>	99,40%	99,30%	99,80%	99,80%	Fontaine progeroid syndrome, 612289
<i>SLC2A1</i>	92,80%	92,80%	100%	100%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
<i>SLC33A1</i>	99,90%	98,90%	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
<i>SLC35A1</i>	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type II f, 603585
<i>SLC35A2</i>	99,90%	98,40%	100%	100%	Congenital disorder of glycosylation, type II m, 300896
<i>SLC35A3</i>	80,70%	78,60%	81,10%	81,00%	?Arthrogyriposis, mental retardation, and seizures, 615553
<i>SLC35C1</i>	99,90%	98,70%	100%	100%	Congenital disorder of glycosylation, type II c, 266265

<i>SLC39A14</i>	100%	99,40%	93,50%	93,50%	?Hyperostosis cranialis interna, 144755 Hypermanganesemia with dystonia 2, 617013
<i>SLC39A8</i>	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type II n, 616721
<i>SLC46A1</i>	99,90%	98,50%	100%	100%	Folate malabsorption, hereditary, 229050
<i>SLC4A4</i>	99,80%	99,20%	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
<i>SLC6A1</i>	100%	100%	100%	100%	Myoclonic-atonic epilepsy, 616421
<i>SLC6A17</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 48, 616269
<i>SLC6A19</i>	100%	100%	100%	100%	Iminoglycinuria, digenic, 242600 HEARTnup disorder, 234500 Hyperglycinuria, 138500
<i>SLC6A3</i>	100%	100%	100%	100%	Parkinsonism-dystonia, infantile, 1, 613135
<i>SLC6A8</i>	93,50%	81,60%	100%	99,80%	Cerebral creatine deficiency syndrome 1, 300352
<i>SLC6A9</i>	100%	100%	100%	100%	Glycine encephalopathy with normal serum glycine, 617301
<i>SLC7A7</i>	100%	99,90%	100%	100%	Lysinuric protein intolerance, 222700
<i>SLC9A6</i>	99,30%	95,50%	100%	98,30%	Mental retardation, X-linked syndromic, Christianson type, 300243
<i>SLC9A7</i>	97,60%	90,30%	99,90%	99,50%	Intellectual developmental disorder, X-linked 108, 301024
<i>SMAD4</i>	100%	99,90%	100%	100%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
<i>SMARCA1</i>	99,60%	97,50%	100%	99,60%	No OMIM disease ID
<i>SMARCA2</i>	96,70%	96,20%	97,40%	96,80%	Nicolaides-Baraitser syndrome, 601358
<i>SMARCA4</i>	99,90%	99,00%	100%	100%	Coffin-Siris syndrome 4, 614609
<i>SMARCB1</i>	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
<i>SMARCC2</i>	99,00%	96,60%	100%	100%	Coffin-Siris syndrome 8, 618362
<i>SMARCD1</i>	94,20%	89,30%	100%	100%	Coffin-Siris syndrome 11, 618779
<i>SMARCE1</i>	96,10%	88,10%	100%	100%	Coffin-Siris syndrome 5, 616938
<i>SMC1A</i>	100%	98,70%	100%	99,80%	Cornelia de Lange syndrome 2, 300590 Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044
<i>SMC3</i>	95,20%	91,00%	100%	100%	Cornelia de Lange syndrome 3, 610759
<i>SMG9</i>	100%	100%	100%	100%	Heart and brain malformation syndrome, 616920
<i>SMOC1</i>	99,90%	98,40%	100%	100%	Microphthalmia with limb anomalies, 206920
<i>SMPD1</i>	100%	100%	100%	100%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
<i>SMPD4</i>	99,40%	94,20%	100%	100%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
<i>SMS</i>	91,50%	78,50%	100%	99,90%	Mental retardation, X-linked, Snyder-Robinson type, 309583

<i>SNAP25</i>	100%	99,90%	100%	100%	?Myasthenic syndrome, congenital, 18, 616330
<i>SNAP29</i>	100%	100%	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
<i>SNIP1</i>	98,90%	97,10%	100%	100%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
<i>SNORD118</i>	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
<i>SNRPB</i>	100%	99,30%	100%	100%	Cerebrocostomandibular syndrome, 117650
<i>SNRPN</i>	100%	97,00%	100%	100%	Prader-Willi syndrome, 176270
<i>SNX14</i>	99,60%	95,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 20, 616354
<i>SOBP</i>	97,50%	92,90%	97,00%	95,30%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
<i>SON</i>	98,80%	94,90%	100%	100%	ZTTK syndrome, 617140
<i>SOS1</i>	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
<i>SOS2</i>	100%	99,20%	100%	100%	Noonan syndrome 9, 616559
<i>SOX10</i>	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
<i>SOX11</i>	100%	100%	100%	100%	Coffin-Siris syndrome 9, 615866
<i>SOX2</i>	100%	100%	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
<i>SOX3</i>	91,40%	75,20%	100%	99,50%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
<i>SOX4</i>	97,30%	90,90%	99,90%	98,20%	Coffin-Siris syndrome 10, 618506
<i>SOX5</i>	99,90%	98,90%	100%	100%	Lamb-Shaffer syndrome, 616803
<i>SPART</i>	99,70%	96,80%	100%	100%	Troyer syndrome, 275900
<i>SPAST</i>	99,80%	98,70%	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
<i>SPATA5</i>	100%	99,70%	100%	100%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
<i>SPECC1L</i>	100%	99,60%	100%	100%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
<i>SPG11</i>	100%	99,30%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
<i>SPOCK1</i>	100%	99,50%	100%	100%	No OMIM disease ID
<i>SPOP</i>	100%	100%	100%	100%	Neurodevelopmental disorder with relative macrocephaly and with or without cardiac or endocrine anomalies, 618829 Neurodevelopmental disorder with microcephaly and dysmorphic facies, 618828
<i>SPR</i>	99,80%	96,30%	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
<i>SPRED1</i>	100%	98,90%	100%	100%	Legius syndrome, 611431

<i>SPTAN1</i>	99,10%	98,60%	100%	100%	Epileptic encephalopathy, early infantile, 5, 613477
<i>SPTBN2</i>	100%	99,30%	99,90%	99,90%	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
<i>SRCAP</i>	99,40%	98,90%	100%	100%	Floating-Harbor syndrome, 136140
<i>SRD5A3</i>	99,90%	99,10%	100%	100%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
<i>SRPX2</i>	99,80%	96,50%	100%	100%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
<i>SSR4</i>	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type Iy, 300934
<i>ST3GAL3</i>	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
<i>ST3GAL5</i>	85,00%	84,20%	98,70%	98,40%	Salt and pepper developmental regression syndrome, 609056
<i>STAG1</i>	99,60%	97,30%	100%	100%	Mental retardation, autosomal dominant 47, 617635
<i>STAG2</i>	97,60%	89,40%	99,90%	98,70%	Mullegama-Klein-Martinez syndrome, 301022 Holoprosencephaly 13, X-linked, 301043
<i>STAMBP</i>	100%	99,40%	100%	100%	Microcephaly-capillary malformation syndrome, 614261
<i>STIL</i>	100%	99,80%	100%	100%	Microcephaly 7, primary, autosomal recessive, 612703
<i>STRA6</i>	100%	99,80%	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
<i>STRADA</i>	100%	98,90%	100%	100%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
<i>STT3A</i>	100%	100%	100%	100%	?Congenital disorder of glycosylation, type Iw, 615596
<i>STT3B</i>	100%	99,60%	100%	100%	?Congenital disorder of glycosylation, type Ix, 615597
<i>STX1B</i>	100%	100%	100%	100%	Generalized epilepsy with febrile seizures plus, type 9, 616172
<i>STXBP1</i>	96,80%	96,50%	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
<i>SUCLA2</i>	94,30%	86,60%	100%	100%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
<i>SUCLG1</i>	99,90%	99,80%	100%	100%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
<i>SUMF1</i>	97,50%	90,80%	100%	100%	Multiple sulfatase deficiency, 272200
<i>SUOX</i>	100%	100%	100%	100%	Sulfite oxidase deficiency, 272300
<i>SUPT16H</i>	98,60%	93,60%	100%	100%	No OMIM disease ID
<i>SURF1</i>	89,40%	88,20%	100%	100%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
<i>SUZ12</i>	91,40%	86,20%	100%	100%	Imagawa-Matsumoto syndrome, 618786
<i>SVBP</i>	100%	100%	100%	100%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
<i>SYN1</i>	81,90%	73,20%	100%	99,60%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
<i>SYNCRIP</i>	97,80%	87,20%	100%	100%	No OMIM disease ID
<i>SYNGAP1</i>	99,40%	98,10%	100%	100%	Mental retardation, autosomal dominant 5, 612621

<i>SYNJ1</i>	99,90%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
<i>SYP</i>	99,90%	96,70%	100%	100%	Mental retardation, X-linked 96, 300802
<i>SYT1</i>	99,80%	98,50%	100%	100%	Baker-Gordon syndrome, 618218
<i>SZT2</i>	99,60%	99,50%	100%	99,90%	Epileptic encephalopathy, early infantile, 18, 615476
<i>TBC1D24</i>	100%	100%	100%	100%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
<i>TBC1D7</i>	100%	99,30%	100%	100%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
<i>TBCD</i>	96,20%	94,40%	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
<i>TBCE</i>	99,80%	97,30%	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
<i>TBCK</i>	99,10%	96,80%	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
<i>TBL1XR1</i>	96,50%	84,90%	100%	100%	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
<i>TBP</i>	100%	99,90%	100%	100%	Spinocerebellar ataxia 17, 607136
<i>TBR1</i>	99,90%	97,90%	100%	100%	Intellectual developmental disorder with autism and speech delay, 606053
<i>TBX1</i>	86,90%	79,50%	94,10%	90,80%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
<i>TCF20</i>	100%	100%	100%	100%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
<i>TCF4</i>	100%	99,80%	100%	100%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
<i>TCF7L2</i>	99,90%	98,80%	100%	100%	No OMIM disease ID
<i>TCN2</i>	100%	100%	100%	100%	Transcobalamin II deficiency, 275350
<i>TCTN2</i>	100%	99,50%	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
<i>TCTN3</i>	100%	100%	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
<i>TDP2</i>	100%	99,40%	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
<i>TECPR2</i>	100%	100%	100%	100%	Spastic paraplegia 49, autosomal recessive, 615031
<i>TECR</i>	100%	98,90%	100%	100%	Mental retardation, autosomal recessive 14, 614020
<i>TELO2</i>	99,70%	96,20%	100%	100%	You-Hoover-Fong syndrome, 616954



<i>TENM3</i>	100%	99,70%	100%	100%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
<i>TET3</i>	94,40%	94,40%	100%	100%	Beck-Fahrner syndrome, 618798
<i>TFAP2A</i>	99,40%	94,30%	100%	100%	Branchiooculofacial syndrome, 113620
<i>TGDS</i>	99,40%	96,80%	100%	100%	Catel-Manzke syndrome, 616145
<i>TGFBR1</i>	93,70%	93,60%	99,00%	96,30%	Loeys-Dietz syndrome 1, 609192
<i>TGIF1</i>	100%	100%	100%	100%	Holoprosencephaly 4, 142946
<i>TH</i>	99,30%	96,10%	100%	100%	Segawa syndrome, recessive, 605407
<i>THOC2</i>	98,80%	93,70%	100%	100%	Mental retardation, X-linked 12/35, 300957
<i>THOC6</i>	100%	100%	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
<i>THRB</i>	100%	99,70%	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
<i>TIMM50</i>	98,30%	94,40%	100%	100%	3-methylglutaconic aciduria, type IX, 617698
<i>TIMM8A</i>	98,00%	90,10%	100%	100%	Mohr-Tranebjaerg syndrome, 304700
<i>TINF2</i>	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
<i>TKFC</i>	100%	99,50%	100%	100%	Triokinase and FMN cyclase deficiency syndrome, 618805
<i>TKT</i>	98,70%	97,80%	98,70%	98,70%	Short stature, developmental delay, and congenital heart defects, 617044
<i>TLK2</i>	99,10%	95,10%	100%	100%	Mental retardation, autosomal dominant 57, 618050
<i>TMCO1</i>	88,00%	87,40%	88,00%	88,00%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
<i>TMEM106B</i>	99,90%	98,80%	100%	100%	Leukodystrophy, hypomyelinating, 16, 617964
<i>TMEM165</i>	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
<i>TMEM216</i>	99,90%	98,10%	100%	100%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
<i>TMEM231</i>	100%	99,60%	100%	100%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
<i>TMEM237</i>	100%	99,90%	100%	100%	Joubert syndrome 14, 614424
<i>TMEM240</i>	100%	100%	100%	100%	Spinocerebellar ataxia 21, 607454
<i>TMEM5</i>	99,50%	96,80%	100%	99,90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
<i>TMEM63A</i>	100%	99,90%	100%	100%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
<i>TMEM67</i>	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
<i>TMEM70</i>	98,00%	93,90%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052

<i>TMLHE</i>	99,50%	97,10%	100%	99,90%	No OMIM disease ID
<i>TMTC3</i>	99,60%	96,50%	100%	100%	Lissencephaly 8, 617255
<i>TMX2</i>	100%	99,80%	100%	100%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
<i>TNIK</i>	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 54, 617028
<i>TNRC6B</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>TOE1</i>	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
<i>TP53RK</i>	92,50%	79,60%	100%	100%	Galloway-Mowat syndrome 4, 617730
<i>TPI1</i>	99,80%	97,50%	100%	100%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
<i>TPO</i>	99,90%	98,20%	100%	100%	Thyroid dysmorphogenesis 2A, 274500
<i>TPP1</i>	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
<i>TPRKB</i>	81,10%	75,90%	81,90%	81,90%	Galloway-Mowat syndrome 5, 617731
<i>TRAF7</i>	100%	99,80%	100%	100%	Cardiac, facial, and digital anomalies with developmental delay, 618164
<i>TRAIP</i>	100%	100%	100%	100%	Seckel syndrome 9, 616777
<i>TRAPPC11</i>	100%	99,20%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
<i>TRAPPC6B</i>	99,90%	98,00%	100%	100%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
<i>TRAPPC9</i>	100%	99,60%	100%	100%	Mental retardation, autosomal recessive 13, 613192
<i>TREX1</i>	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
<i>TRIM32</i>	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
<i>TRIM8</i>	99,90%	99,10%	100%	100%	No OMIM disease ID
<i>TRIO</i>	99,20%	97,50%	99,30%	98,40%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
<i>TRIP12</i>	99,90%	99,20%	100%	100%	Mental retardation, autosomal dominant 49, 617752
<i>TRIT1</i>	100%	100%	100%	100%	Combined oxidative phosphorylation deficiency 35, 617873
<i>TRMT1</i>	99,40%	96,20%	100%	100%	Mental retardation, autosomal recessive 68, 618302
<i>TRMT10A</i>	100%	99,70%	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
<i>TRNT1</i>	99,50%	96,50%	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
<i>TRPM3</i>	100%	99,50%	100%	100%	No OMIM disease ID
<i>TRRAP</i>	99,90%	99,50%	100%	100%	Developmental delay with or without dysmorphic facies and autism, 618454 ?Deafness, autosomal dominant 75, 618778
<i>TSC1</i>	99,80%	98,70%	100%	100%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangiomyomatosis, 606690

<i>TSC2</i>	100%	99,60%	100%	100%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangiomyomatosis, somatic, 606690
<i>TSEN15</i>	99,80%	97,50%	100%	100%	Pontocerebellar hypoplasia, type 2F, 617026
<i>TSEN2</i>	100%	99,60%	100%	100%	Pontocerebellar hypoplasia type 2B, 612389
<i>TSEN54</i>	96,30%	94,30%	99,90%	98,90%	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
<i>TSFM</i>	100%	99,50%	94,90%	94,90%	Combined oxidative phosphorylation deficiency 3, 610505
<i>TSHB</i>	100%	100%	100%	100%	Hypothyroidism, congenital, nongoitrous 4, 275100
<i>TSPAN7</i>	100%	100%	100%	100%	Mental retardation, X-linked 58, 300210
<i>TTC19</i>	81,50%	73,80%	100%	99,20%	Mitochondrial complex III deficiency, nuclear type 2, 615157
<i>TTC37</i>	100%	99,30%	100%	100%	Trichohepatoenteric syndrome 1, 222470
<i>TTC8</i>	99,60%	98,10%	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
<i>TTI2</i>	100%	100%	100%	100%	Mental retardation, autosomal recessive 39, 615541
<i>TUBA1A</i>	99,90%	97,00%	100%	100%	Lissencephaly 3, 611603
<i>TUBA8</i>	99,90%	99,50%	100%	100%	Cortical dysplasia, complex, with other brain malformations 8, 613180
<i>TUBB</i>	97,30%	93,90%	99,80%	99,80%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
<i>TUBB2A</i>	97,00%	95,70%	100%	100%	Cortical dysplasia, complex, with other brain malformations 5, 615763
<i>TUBB2B</i>	100%	99,50%	100%	100%	Cortical dysplasia, complex, with other brain malformations 7, 610031
<i>TUBB3</i>	98,30%	96,90%	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
<i>TUBB4A</i>	95,90%	94,00%	97,10%	96,00%	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
<i>TUBG1</i>	100%	100%	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
<i>TUBGCP2</i>	99,70%	96,20%	97,00%	97,00%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
<i>TUBGCP4</i>	99,20%	96,40%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
<i>TUBGCP6</i>	100%	99,30%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
<i>TUSC3</i>	100%	99,50%	100%	100%	Mental retardation, autosomal recessive 7, 611093
<i>TWIST1</i>	100%	98,90%	97,20%	92,30%	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
<i>TWNK</i>	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138

UBA5	97,80%	86,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	99,70%	96,00%	100%	99,70%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	99,10%	94,80%	100%	100%	Angelman syndrome, 105830
UBE3B	100%	99,90%	100%	100%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	99,90%	99,10%	98,00%	98,00%	Johanson-Blizzard syndrome, 243800
UBTF	100%	99,40%	100%	100%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	100%	100%	100%	100%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	74,00%	69,40%	100%	100%	Leukodystrophy, hypomyelinating, 14, 617899
UGDH	99,90%	99,10%	100%	100%	Epileptic encephalopathy, early infantile, 84, 618792
UGP2	99,00%	98,60%	96,30%	96,30%	Epileptic encephalopathy, early infantile, 83, 618744
UNC13A	99,30%	97,70%	100%	100%	No OMIM disease ID
UNC80	100%	99,50%	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	100%	100%	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	92,20%	84,10%	100%	100%	Mental retardation, X-linked, syndromic 14, 300676
UROC1	100%	100%	100%	100%	?Urocanase deficiency, 276880
USP27X	100%	100%	100%	100%	Mental retardation, X-linked 105, 300984
USP7	96,30%	92,70%	100%	100%	No OMIM disease ID
USP9X	98,20%	92,90%	100%	100%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
VAMP1	100%	100%	100%	100%	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VAMP2	99,50%	97,70%	100%	100%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VARS	100%	99,90%	100%	100%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VLDLR	100%	99,80%	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	94,90%	93,60%	100%	100%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	99,50%	98,20%	99,50%	99,40%	Cohen syndrome, 216550
VPS37A	91,30%	78,20%	100%	100%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	91,50%	90,70%	100%	99,30%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,70%	98,50%	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	100%	99,70%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	100%	99,70%	100%	100%	Desanto-Shinawi syndrome, 616708
WARS2	100%	99,40%	100%	100%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	99,90%	96,50%	100%	100%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	99,10%	95,50%	100%	100%	?Mental retardation, autosomal recessive 43, 615817

WDFY3	100%	99,60%	100%	100%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	98,20%	94,40%	98,10%	98,10%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR13	99,90%	98,60%	100%	100%	No OMIM disease ID
WDR26	99,00%	96,60%	100%	100%	Skraban-Deardorff syndrome, 617616
WDR37	100%	99,60%	100%	100%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100%	100%	100%	100%	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45	96,40%	89,70%	100%	100%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	98,00%	89,20%	100%	100%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	100%	99,50%	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%	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	100%	99,90%	100%	100%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WWOX	100%	100%	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211
XPA	99,60%	95,60%	100%	100%	Xeroderma pigmentosum, group A, 278700
XRCC4	99,90%	99,30%	100%	100%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,40%	89,60%	98,10%	94,80%	Desbuquois dysplasia 2, 615777
YME1L1	99,00%	95,20%	100%	100%	?Optic atrophy 11, 617302
YWHAE	100%	100%	100%	100%	No OMIM disease ID
YWHAG	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 56, 617665
YY1	100%	99,80%	100%	100%	Gabriele-de Vries syndrome, 617557
ZBTB11	99,90%	99,60%	100%	100%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100%	99,90%	100%	100%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZBTB18	100%	99,90%	100%	99,80%	Mental retardation, autosomal dominant 22, 612337
ZBTB20	100%	100%	100%	100%	PriINTELLECTUAL DISABILITYose syndrome, 259050
ZBTB24	100%	100%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	99,90%	98,90%	100%	100%	Mental retardation, autosomal recessive 56, 617125

ZC4H2	100%	99,00%	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZDHHC9	99,90%	93,80%	100%	100%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	99,90%	99,10%	97,40%	97,40%	Mowat-Wilson syndrome, 235730
ZFYVE26	100%	99,10%	100%	100%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	100%	100%	100%	100%	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 ?Craniosynostosis 6, 616602
ZIC2	100%	98,70%	98,50%	95,70%	Holoprosencephaly 5, 609637
ZMIZ1	99,40%	98,40%	100%	100%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYND11	100%	99,60%	100%	100%	Mental retardation, autosomal dominant 30, 616083
ZNF142	100%	99,90%	100%	100%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	99,90%	99,60%	100%	100%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99,60%	98,30%	99,60%	99,60%	No OMIM disease ID
ZNF335	100%	99,90%	100%	100%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	99,90%	99,30%	100%	100%	No OMIM disease ID
ZNF41	100%	99,60%	100%	100%	No OMIM disease ID
ZNF462	100%	99,90%	100%	100%	Weiss-Kruszka syndrome, 618619
ZNF711	99,80%	98,20%	100%	100%	Mental retardation, X-linked 97, 300803
ZSWIM6	95,50%	91,90%	94,90%	92,10%	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

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

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-DNA coding genes.

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

OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.

This list is accurate for panel version DG 2.18

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