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, 1O, 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

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

ALDH4A1	100%	99,40%	100%	100%	Hyperprolinemia, type II, 239510
ALDH5A1	91,00%	81,50%	100%	100%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	94,40%	88,80%	100%	100%	Epilepsy, pyridoxine-dependent, 266100
ALG1	53,00%	45,80%	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,80%	96,80%	96,80%	96,80%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	98,40%	92,60%	100%	99,60%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG2	100%	100%	100%	100%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,60%	94,80%	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	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
ALG9	100%	99,70%	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	99,80%	98,90%	100%	100%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
ALX3	77,90%	73,30%	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	99,30%	100%	100%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMER1	99,90%	98,50%	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100%	99,10%	100%	100%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	99,80%	98,90%	100%	100%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100%	100%	100%	100%	Glycine encephalopathy, 605899
ANK3	99,30%	99,00%	100%	100%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	100%	100%	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	99,90%	98,60%	100%	99,80%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	97,50%	94,80%	100%	100%	KBG syndrome, 148050
ANKS1B	100%	99,60%	100%	100%	No OMIM disease ID
ANO10	99,80%	97,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	99,70%	97,90%	100%	100%	GAPO syndrome, 230740
AP1S1	99,90%	99,50%	100%	100%	MEDNIK syndrome, 609313
AP1S2	76,40%	67,90%	100%	100%	Mental retardation, X-linked syndromic 5, 304340
AP2M1	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
ATP6V0A2	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

ATP7A	99,70%	97,50%	100%	100%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	100%	99,70%	100%	100%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATR	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	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
AUH	100%	99,80%	100%	100%	3-methylglutaconic aciduria, type I, 250950
AUTS2	98,20%	95,80%	100%	100%	Mental retardation, autosomal dominant 26, 615834
AVPR2	100%	99,40%	100%	100%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
B3GALNT2	93,80%	89,40%	92,50%	92,50%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	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
B3GLCT	99,60%	96,30%	99,90%	99,20%	Peters-plus syndrome, 261540
B4GALNT1	99,30%	95,00%	100%	100%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT7	99,80%	97,40%	99,90%	98,60%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAZ2B	99,90%	99,00%	100%	100%	No OMIM disease ID
BBS1	100%	100%	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	99,80%	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	100%	99,50%	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,90%	99,30%	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	99,00%	93,90%	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	98,70%	95,50%	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	99,70%	97,60%	100%	100%	Bardet-Biedl syndrome 9, 615986
BCAP31	92,60%	83,20%	100%	99,90%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	99,90%	99,20%	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,50%	94,40%	100%	100%	Maple syrup urine disease, type Ib, 248600
BCKDK	100%	100%	100%	100%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL11A	99,80%	98,10%	100%	100%	Dias-Logan syndrome, 617101

BCL11B	99,10%	95,60%	98,80%	97,30%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCOR	99,60%	97,40%	100%	99,90%	Microphthalmia, syndromic 2, 300166
BCORL1	99,60%	97,90%	100%	100%	Shukla-Vernon syndrome, 301029
BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	99,80%	98,30%	100%	100%	Bloom syndrome, 210900
BOLA3	99,40%	90,20%	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	96,10%	94,20%	99,60%	98,60%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	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
BRAT1	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
BRF1	99,90%	98,40%	100%	100%	Cerebellofaciodental syndrome, 616202
BRPF1	100%	100%	100%	100%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	99,50%	97,40%	100%	100%	No OMIM disease ID
BRWD3	99,30%	97,20%	100%	100%	Mental retardation, X-linked 93, 300659
BSCL2	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
BTD	100%	99,90%	100%	100%	Biotinidase deficiency, 253260
BUB1B	99,60%	98,90%	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
C12orf4	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 66, 618221
C12orf57	100%	98,90%	100%	100%	Temtamy syndrome, 218340
C12orf65	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C2CD3	95,80%	95,60%	95,90%	95,90%	Orofaciodigital syndrome XIV, 615948
C5orf42	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

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

CEP41	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
CEP57	99,20%	93,00%	100%	100%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP83	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
CEP89	96,00%	94,50%	100%	100%	No OMIM disease ID
CHAMP1	100%	100%	100%	100%	Mental retardation, autosomal dominant 40, 616579
CHD1	99,30%	94,90%	100%	100%	Pilarowski-Bjornsson syndrome, 617682
CHD2	99,40%	99,20%	100%	100%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	94,80%	92,60%	99,80%	99,50%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	99,90%	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	100%	99,90%	100%	100%	No OMIM disease ID
CHKB	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CHRNA4	98,30%	96,20%	100%	100%	Epilepsy, nocturnal frontal lobe, 1, 600513
CIC	63,30%	63,30%	100%	99,90%	Mental retardation, autosomal dominant 45, 617600
CIT	100%	99,40%	100%	100%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	99,70%	98,60%	100%	100%	Filippi syndrome, 272440
CLCN4	99,90%	98,90%	100%	100%	Raynaud-Claes syndrome, 300114
CLIC2	99,90%	96,50%	100%	100%	?Mental retardation, X-linked, syndromic 32, 300886
CLIP1	100%	99,00%	100%	100%	No OMIM disease ID
CLN3	92,50%	91,80%	92,50%	92,50%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	99,40%	95,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,90%	97,10%	100%	100%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,50%	83,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLTC	100%	99,90%	100%	100%	Mental retardation, autosomal dominant 56, 617854
CNKSR2	98,60%	93,60%	100%	100%	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	100%	100%	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNOT1	100%	99,90%	100%	100%	Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	99,90%	99,50%	100%	100%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100%	100%	100%	100%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672

CNPY3	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 60, 617929
CNTNAP2	100%	99,80%	100%	100%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COASY	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COG1	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIg, 611209
COG4	100%	99,90%	100%	100%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	99,70%	97,60%	100%	100%	Congenital disorder of glycosylation, type IIi, 613612
COG6	99,10%	93,90%	100%	100%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	99,60%	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	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
COL4A2	100%	99,60%	100%	100%	Brain small vessel disease 2, 614483
COL4A3BP	99,60%	96,50%	100%	100%	Mental retardation, autosomal dominant 34, 616351
COLEC11	100%	100%	100%	100%	3MC syndrome 2, 265050
COQ2	98,00%	95,30%	97,20%	97,20%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,90%	89,30%	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100%	99,50%	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	100%	97,90%	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX15	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
COX6B1	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110
CPLX1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 63, 617976
CPS1	100%	99,90%	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300
CRADD	99,50%	96,30%	100%	100%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	88,20%	87,70%	97,00%	92,90%	Mental retardation, autosomal recessive 2, 607417
CREBBP	99,70%	98,50%	100%	100%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRLF1	91,00%	89,80%	97,90%	95,20%	Cold-induced sweating syndrome 1, 272430
CSDE1	99,90%	99,50%	100%	100%	No OMIM disease ID

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

DARS	100%	99,30%	100%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	99,30%	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	99,80%	98,00%	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	98,90%	93,30%	100%	100%	Woodhouse-Sakati syndrome, 241080
DCC	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
DCHS1	99,80%	99,10%	100%	100%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	100%	100%	100%	100%	Al-Raqad syndrome, 616459
DCX	100%	99,90%	100%	100%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDC	99,70%	96,40%	100%	100%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	100%	99,60%	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	85,20%	80,70%	100%	100%	Warsaw breakage syndrome, 613398
DDX3X	86,80%	84,90%	100%	100%	Mental retardation, X-linked 102, 300958
DDX59	100%	100%	100%	100%	Orofaciodigital syndrome V, 174300
DDX6	97,70%	88,70%	100%	100%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	97,30%	88,80%	100%	98,70%	Mental retardation, autosomal dominant 24, 615828 ?Dyskinesia, seizures, and intellectual developmental disorder, 617171
DEGS1	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	100%	99,80%	100%	100%	Epilepsy, familial focal, with variable foci 1, 604364
DHCR24	100%	100%	100%	100%	Desmosterolosis, 602398
DHCR7	100%	100%	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	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
DHFR	92,10%	78,90%	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	100%	99,70%	93,30%	93,20%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	99,90%	98,90%	100%	100%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	100%	99,90%	100%	100%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DIAPH1	99,80%	99,00%	99,50%	97,90%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIP2B	100%	99,30%	100%	100%	Mental retardation, FRA12A type, 136630

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

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

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

FGF12	99,90%	98,10%	100%	100%	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	100%	100%	100%	100%	Spinocerebellar ataxia 27, 609307
FGFR1	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
FGFR2	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
FGFR3	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

FH	92,10%	88,30%	100%	100%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	100%	100%	100%	100%	Thauvin-Robinet-Faivre syndrome, 617107
FIGN	100%	100%	100%	100%	No OMIM disease ID
FMR 1	96,10%	92,10%	100%	100%	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624
FKRP	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
FKTN	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
FLNA	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
FLVCR1	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100%	100%	100%	100%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	85,50%	82,50%	100%	100%	Mental retardation, autosomal recessive 47, 616193
FOLR1	100%	100%	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	88,60%	82,10%	99,20%	96,40%	Rett syndrome, congenital variant, 613454
FOXJ1	99,90%	98,40%	100%	100%	Ciliary dyskinesia, primary, 43, 618699
FOXP1	100%	99,80%	100%	100%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	99,50%	99,20%	100%	100%	Speech-language disorder-1, 602081
FOXRED1	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	100%	99,40%	100%	100%	Fraser syndrome 1, 219000
FRMD4A	90,70%	87,30%	96,60%	96,60%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMPD4	99,80%	98,50%	100%	100%	Mental retardation, X-linked 104, 300983
FRRS1L	79,70%	69,10%	99,20%	95,80%	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	95,70%	91,00%	100%	100%	Glutamate formiminotransferase deficiency, 229100

FTO	83,80%	83,70%	94,20%	94,20%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	98,00%	93,80%	100%	100%	Mental retardation, X-linked 9/44, 309549
FUCA1	100%	99,90%	100%	100%	Fucosidosis, 230000
FUT8	100%	99,20%	100%	100%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
GABBR2	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
GABRA1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744
GABRA3	98,70%	94,40%	99,90%	98,90%	No OMIM disease ID
GABRB1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	100%	99,90%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	99,60%	98,20%	100%	100%	Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	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
GAD1	100%	99,90%	100%	100%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	99,80%	98,30%	100%	100%	Krabbe disease, 245200
GALE	100%	100%	100%	100%	Galactose epimerase deficiency, 230350
GALT	100%	99,70%	100%	100%	Galactosemia, 230400
GAMT	93,10%	82,70%	100%	100%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	100%	100%	100%	100%	Mental retardation, autosomal dominant 18, 615074
GATM	100%	100%	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	99,90%	95,50%	100%	100%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCSH	75,70%	68,90%	100%	100%	?Glycine encephalopathy, 605899
GDI1	99,80%	98,70%	100%	100%	Mental retardation, X-linked 41, 300849
GFAP	91,80%	89,70%	100%	100%	Alexander disease, 203450
GFM1	99,90%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,90%	95,20%	100%	100%	Combined oxidative phosphorylation deficiency 39, 618397
GJA1	100%	100%	100%	100%	Erythrokeratodermia 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
GJB1	100%	100%	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800

GJC2	78,20%	58,70%	96,90%	91,40%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GK	88,90%	70,40%	100%	99,90%	Glycerol kinase deficiency, 307030
GLB1	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
GLDC	89,90%	82,00%	100%	99,90%	Glycine encephalopathy, 605899
GLI2	99,10%	97,40%	100%	99,80%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	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
GLIS3	100%	99,60%	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLUD1	94,20%	82,90%	100%	100%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLYCTK	98,80%	97,30%	100%	100%	D-glyceric aciduria, 220120
GM2A	100%	100%	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPA	100%	100%	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (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
GNAO1	93,80%	93,80%	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAS	100%	99,90%	100%	99,90%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GNB1	100%	100%	100%	100%	Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GNB5	100%	98,80%	100%	100%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	99,70%	97,30%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765

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

HADH	99,00%	97,50%	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	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
HAX1	100%	100%	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	99,80%	97,60%	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	98,30%	93,60%	100%	100%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	100%	99,70%	100%	100%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	100%	99,80%	100%	100%	No OMIM disease ID
HDAC6	99,50%	97,40%	100%	100%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	100%	99,80%	100%	100%	Cornelia de Lange syndrome 5, 300882
HECW2	100%	99,10%	100%	100%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	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
HERC1	100%	100%	100%	100%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	79,90%	77,20%	100%	100%	Mental retardation, autosomal recessive 38, 615516
HESX1	99,70%	97,30%	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	93,80%	93,30%	100%	100%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	99,60%	96,90%	100%	99,90%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	86,40%	86,30%	91,20%	89,30%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	98,20%	88,50%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIST1H1E	100%	100%	100%	100%	Rahman syndrome, 617537
HIST1H4C	100%	100%	100%	100%	No OMIM disease ID
HIVEP2	100%	100%	100%	100%	Mental retardation, autosomal dominant 43, 616977
HLCS	100%	100%	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMGCL	100%	99,80%	100%	100%	HMG-CoA lyase deficiency, 246450
HNMT	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 51, 616739
HNRNPH2	100%	100%	100%	100%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	91,50%	82,80%	100%	100%	Au-Kline syndrome, 616580

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

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

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

KIDINS220	100%	100%	100%	100%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	97,60%	94,80%	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	99,60%	97,70%	100%	100%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	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
KIF1BP	96,10%	96,10%	96,10%	96,10%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF2A	99,60%	95,60%	100%	100%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	99,40%	95,70%	100%	100%	?Mental retardation, X-linked 100, 300923
KIF5C	99,90%	98,80%	99,80%	99,80%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	93,60%	90,60%	99,10%	97,80%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL3	99,80%	98,90%	100%	100%	No OMIM disease ID
KLF7	100%	99,70%	100%	100%	No OMIM disease ID
KLHL15	100%	99,70%	100%	100%	Mental retardation, X-linked 103, 300982
KMT2A	100%	99,90%	99,90%	99,40%	Wiedemann-Steiner syndrome, 605130
KMT2B	95,80%	94,00%	98,70%	97,90%	Dystonia 28, childhood-onset, 617284
KMT2C	92,20%	91,00%	100%	100%	Kleefstra syndrome 2, 617768
KMT2D	100%	99,40%	100%	100%	Kabuki syndrome 1, 147920
KMT2E	99,80%	98,50%	100%	100%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	99,90%	99,10%	100%	100%	Mental retardation, autosomal dominant 51, 617788
KNL1	99,20%	98,10%	98,90%	98,80%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100%	100%	100%	100%	Mental retardation, autosomal recessive 41, 615637
KRAS	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
L1CAM	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
L2HGDH	99,00%	97,20%	100%	100%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	100%	99,70%	100%	100%	Poretti-Boltshauser syndrome, 615960
LAMA2	100%	99,60%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMB1	100%	99,90%	100%	100%	Lissencephaly 5, 615191
LAMC3	98,60%	97,10%	100%	99,60%	Cortical malformations, occipital, 614115
LAMP2	99,20%	95,60%	100%	100%	Danon disease, 300257
LARGE1	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
LARP7	88,50%	78,40%	100%	100%	Alazami syndrome, 615071
LAS1L	99,70%	97,30%	100%	100%	Wilson-Turner syndrome, 309585
LIAS	100%	99,10%	100%	100%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	100%	99,90%	100%	100%	LIG4 syndrome, 606593
LINGO1	100%	100%	100%	100%	Mental retardation, autosomal recessive 64, 618103
LINS1	100%	99,10%	100%	100%	Mental retardation, autosomal recessive 27, 614340
LMAN2L	100%	99,70%	100%	100%	?Mental retardation, autosomal recessive, 52, 616887
LONP1	100%	99,80%	100%	100%	CODAS syndrome, 600373
LRP2	100%	99,90%	100%	100%	Donnai-Barrow syndrome, 222448
LRPPRC	99,90%	99,10%	100%	100%	Leigh syndrome, French-Canadian type, 220111
LYST	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
LZTFL1	99,90%	99,20%	100%	100%	Bardet-Biedl syndrome 17, 615994
LZTR1	100%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAB21L1	100%	100%	100%	100%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100%	99,60%	100%	100%	Lissencephaly 9 with complex brainstem malformation, 618325
MAF	83,50%	78,00%	88,60%	82,20%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAG	100%	100%	100%	100%	Spastic paraplegia 75, autosomal recessive, 616680

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

MED13L	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
MED17	96,30%	93,50%	100%	100%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 18, 614249
MED25	100%	99,80%	100%	100%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	99,90%	96,00%	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	99,90%	99,00%	100%	100%	Carpenter syndrome 2, 614976
MEIS2	100%	100%	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
METTL23	100%	100%	100%	100%	Mental retardation, autosomal recessive 44, 615942
METTL5	99,30%	98,50%	99,90%	98,30%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	94,30%	89,90%	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	99,70%	98,50%	100%	100%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	100%	99,70%	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MGP	98,70%	95,10%	100%	100%	Keutel syndrome, 245150
MICU1	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
MID1	99,80%	98,70%	100%	100%	Opitz GBBB syndrome, type I, 300000
MID2	99,80%	98,70%	100%	100%	?Mental retardation, X-linked 101, 300928
MKKS	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	100%	99,00%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	96,00%	90,40%	100%	98,90%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	100%	99,60%	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	100%	100%	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	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
MN1	100%	99,30%	100%	100%	Meningioma, 607174 CEBALID syndrome, 618774
MOCS1	99,20%	95,40%	100%	100%	Molybdenum cofactor deficiency A, 252150

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

NARS2	98,30%	97,40%	100%	100%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAXE	100%	99,80%	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	92,00%	90,60%	100%	100%	No OMIM disease ID
NBN	99,90%	98,60%	100%	100%	Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260 Leukemia, acute lymphoblastic, 613065
NCAPG2	99,90%	99,20%	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NDE1	100%	100%	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDP	100%	99,70%	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	100%	100%	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	99,90%	99,30%	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100%	100%	100%	99,80%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFAF3	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	62,60%	61,10%	100%	99,60%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,70%	90,60%	91,90%	90,70%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100%	99,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	98,00%	96,10%	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,90%	76,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	72,00%	71,50%	100%	100%	Periventricular nodular heterotopia 7, 617201
NEU1	99,70%	97,70%	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	100%	99,50%	100%	100%	Mental retardation, X-linked 98, 300912
NF1	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
NFIA	100%	99,60%	100%	100%	Brain malformations with or without urinary tract defects, 613735
NFIB	97,40%	96,50%	100%	100%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100%	99,50%	99,60%	98,70%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFU1	98,80%	90,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100%	99,80%	100%	100%	Congenital disorder of deglycosylation, 615273
NHS	95,40%	93,90%	100%	99,80%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIPBL	98,90%	97,00%	100%	100%	Cornelia de Lange syndrome 1, 122470
NKAP	99,30%	95,20%	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
NKX2-1	98,60%	85,60%	100%	100%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN3	99,90%	99,00%	100%	100%	No OMIM disease ID
NLGN4X	99,90%	98,90%	100%	99,90%	Mental retardation, X-linked, 300495
NONO	100%	98,40%	100%	100%	Mental retardation, X-linked, syndromic 34, 300967
NOVA2	99,00%	94,60%	96,80%	93,30%	No OMIM disease ID
NPC1	99,60%	98,70%	100%	100%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100%	99,60%	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	100%	99,00%	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NR2F1	100%	100%	99,10%	95,10%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	100%	100%	100%	100%	No OMIM disease ID
NRAS	100%	100%	100%	100%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 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
NRXN1	97,40%	96,90%	100%	99,80%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	100%	99,90%	100%	100%	Sotos syndrome 1, 117550
NSD2	99,90%	99,20%	100%	100%	No OMIM disease ID

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

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

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

PHGDH	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	98,60%	96,10%	100%	99,70%	Chung-Jansen syndrome, 617991
PI4KA	92,60%	88,80%	99,90%	99,90%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
TAF1	99,80%	97,70%	100%	100%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	100%	100%	100%	100%	Mental retardation, autosomal recessive 60, 617432
TAF2	99,90%	98,60%	100%	100%	Mental retardation, autosomal recessive 40, 615599
TAF6	99,80%	98,90%	100%	100%	Alazami-Yuan syndrome, 617126
TANC2	100%	99,50%	100%	100%	No OMIM disease ID
TANGO2	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	99,50%	97,90%	100%	100%	No OMIM disease ID
TASP1	99,70%	98,80%	100%	100%	No OMIM disease ID
TAT	100%	100%	100%	100%	Tyrosinemia, type II, 276600
TBC1D20	94,20%	94,20%	100%	99,90%	Warburg micro syndrome 4, 615663
TBC1D23	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 11, 617695
PIGA	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	99,90%	97,80%	100%	100%	Epileptic encephalopathy, early infantile, 80, 618580
PIGC	99,20%	90,90%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 53, 616917
PIGL	100%	100%	100%	100%	CHIME syndrome, 280000
PIGN	93,80%	91,50%	98,80%	98,80%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,80%	87,30%	100%	100%	Epileptic encephalopathy, early infantile, 55, 617599
PIGT	98,10%	98,10%	100%	100%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100%	99,10%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 21, 618590
PIGV	100%	100%	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100%	99,80%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	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 Macrodactyly, 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
PIK3R2	90,70%	89,60%	99,30%	96,10%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLAA	100%	99,20%	100%	100%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLK4	99,90%	98,20%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	100%	99,20%	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	98,20%	90,10%	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLXND1	98,90%	96,20%	99,70%	99,40%	No OMIM disease ID
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	97,70%	94,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100%	99,70%	100%	100%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	100%	100%	100%	100%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	99,80%	98,90%	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	100%	99,70%	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POGZ	99,40%	99,00%	100%	100%	White-Sutton syndrome, 616364
POLA1	99,30%	95,40%	100%	99,90%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLG	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

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

PPT1	90,30%	90,30%	82,50%	82,50%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	100%	100%	100%	Renpenning syndrome, 309500
PRKAR1A	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
PRMT7	100%	99,90%	100%	100%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	85,00%	80,60%	100%	100%	Hyperprolinemia, type I, 239500
PRPS1	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
PRR12	98,70%	97,20%	100%	100%	No OMIM disease ID
PRSS12	100%	99,90%	100%	100%	Mental retardation, autosomal recessive 1, 249500
PRUNE1	100%	99,90%	100%	100%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PSAP	100%	100%	100%	100%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	95,30%	81,60%	100%	100%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMD12	98,60%	92,90%	100%	100%	Stankiewicz-Isidor syndrome, 617516
PSPH	100%	100%	100%	100%	Phosphoserine phosphatase deficiency, 614023
PTCH1	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCHD1	100%	99,90%	100%	100%	No OMIM disease ID
PTDSS1	100%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	99,50%	97,00%	100%	100%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTF1A	95,80%	85,60%	98,60%	93,30%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTPN11	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250

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

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

RORA	96,70%	90,20%	100%	100%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPGRIP1L	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPL10	97,40%	89,10%	100%	100%	Mental retardation, X-linked, syndromic, 35, 300998
RPS19	100%	99,60%	100%	100%	Diamond-Blackfan anemia 1, 105650
RPS6KA3	98,40%	94,40%	99,90%	98,80%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RRM2B	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
RSPRY1	100%	100%	100%	100%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	99,80%	96,80%	100%	100%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	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
RTN4IP1	99,90%	98,70%	100%	100%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	98,90%	98,00%	100%	100%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,40%	97,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100%	100%	100%	100%	Mental retardation, autosomal recessive 61, 617773
SALL1	99,90%	99,00%	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS	100%	99,30%	100%	100%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SATB2	99,70%	97,40%	100%	100%	Glass syndrome, 612313
SBDS	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
SC5D	100%	99,50%	100%	100%	Lathosterolosis, 607330
SCAMP5	100%	100%	100%	100%	No OMIM disease ID
SCAPER	99,70%	98,20%	100%	100%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCN1A	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
SCN1B	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
SHOC2	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
SHROOM4	100%	99,00%	100%	100%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	98,70%	94,40%	100%	100%	Epileptic encephalopathy, early infantile, 30, 616341
SIL1	99,20%	96,70%	100%	100%	Marinesco-Sjogren syndrome, 248800
SIN3A	100%	99,00%	100%	100%	Witteveen-Kolk syndrome, 613406
SIX3	99,90%	98,60%	100%	100%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	99,30%	94,90%	100%	99,40%	Shprintzen-Goldberg syndrome, 182212
SLC12A5	83,90%	83,80%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 34, 616645
SLC12A6	100%	100%	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	99,20%	93,70%	100%	100%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	99,60%	97,00%	100%	100%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC19A3	100%	99,80%	98,70%	98,70%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	99,90%	99,60%	100%	100%	Dicarboxylic aminoaciduria, 222730
SLC1A2	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	99,00%	95,80%	100%	100%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	95,80%	88,60%	99,50%	97,80%	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	99,90%	99,50%	100%	100%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	99,80%	98,10%	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	98,60%	95,80%	100%	100%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	99,40%	99,30%	99,80%	99,80%	Fontaine progeroid syndrome, 612289
SLC2A1	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
SLC33A1	99,90%	98,90%	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	99,90%	98,40%	100%	100%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	80,70%	78,60%	81,10%	81,00%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	99,90%	98,70%	100%	100%	Congenital disorder of glycosylation, type IIc, 266265

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

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

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

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

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

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

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