INTELLECTUAL DISABILITY GENE PANEL DG 3.1.0 (1392 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS1	100	99,9	100	100	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AASS	100	99,7	100	100	Hyperlysinemia, 238700
ABAT	100	99,4	100	100	GABA-transaminase deficiency, 613163
ABCA2	99,7	98,9	100	100	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCC8	100	99,8	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	100	99,9	100	100	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABCD1	75,8	71,6	100	100	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD4	99,9	98,6	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	100	100	100	100	Chanarin-Dorfman syndrome, 275630
ACAD9	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	99,9	98,2	100	100	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACAT1	99,9	97,6	100	100	Alpha-methylacetoacetic aciduria, 203750
ACO2	96,3	90,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100	99,9	100	100	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	100	99,9	100	100	Combined malonic and methylmalonic aciduria, 614265
ACSL4	98,7	94,6	100	100	Mental retardation, X-linked 63, 300387
ACTB	99,7	96,1	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	99,8	98,7	100	100	No OMIM disease ID
ACTL6B	100	99,8	100	100	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACVR1	100	100	100	100	Fibrodysplasia ossificans progressiva, 135100
ACY1	100	98,8	100	100	Aminoacylase 1 deficiency, 609924
ADAM22	99,9	99,5	100	100	Developmental and epileptic encephalopathy 61, 617933
ADAR	100	99,8	100	100	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADARB1	97,9	95,2	95,1	95,1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100	99,7	100	100	Mental retardation, autosomal recessive 36, 615286
ADGRG1	100	100	100	100	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADK	84,1	81	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	90,5	90,5	95,4	95,4	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100	99,8	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	99,2	98,7	100	100	Adenylosuccinase deficiency, 103050
AFF2	99,9	99,4	100	99,8	Mental retardation, X-linked, FRAXE type, 309548
AFF4	99,9	98,9	100	100	CHOPS syndrome, 616368
AFG3L2	95	91,1	100	99,9	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
AGMO	99,2	96	100	100	No OMIM disease ID
AGO2	99,1	99,1	99,9	99,5	Lessel-Kreienkamp syndrome, 619149
AGTPBP1	96	94,1	100	100	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AHCY	100	99,2	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100	99,3	100	100	Xia-Gibbs syndrome, 615829
AHI1	99,7	97,9	100	100	Joubert syndrome 3, 608629
AHSG	99,9	99,5	100	100	?Alopecia-mental retardation syndrome 1, 203650
AIFM1	99,9	98,8	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	99,2	94,5	100	99,9	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	88,9	86	100	100	Leukodystrophy, hypomyelinating, 17, 618006

AKT3	98,7	94,5	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	100	99,9	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	88,8	88,1	93,2	93,2	Sjogren-Larsson syndrome, 270200
ALDH4A1	100	99,4	100	100	Hyperprolinemia, type II, 239510
ALDH5A1	91	81,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	94,4	88,8	100	100	Epilepsy, pyridoxine-dependent, 266100
ALG1	53	45,8	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100	100	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	98,4	92,6	100	99,6	Developmental and epileptic encephalopathy 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG14	100	99,9	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031
ALG2	100	100	100	100	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,6	94,8	100	100	Congenital disorder of glycosylation, type Ic, 603147
ALG8	97,2	95,6	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100	99,7	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	99,8	98,9	100	100	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ALX3	77,9	73,3	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,3	100	100	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMER1	99,9	98,5	100	100	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100	99,1	100	100	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	99,8	98,9	100	100	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100	100	100	100	Glycine encephalopathy, 605899
ANK3	99,3	99	100	100	Mental retardation, autosomal recessive, 37, 615493

ANKH	100	100	100	100	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKLE2	99,9	98,6	100	99,8	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	96,1	93,5	100	100	KBG syndrome, 148050
ANKS1B	100	99,6	100	100	No OMIM disease ID
ANO10	99,8	97,9	100	100	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	99,7	97,9	100	100	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S1	99,9	99,5	100	100	MEDNIK syndrome, 609313
AP1S2	77,9	69,9	100	100	Mental retardation, X-linked syndromic 5, 304340
AP2M1	100	100	100	100	Intellectual developmental disorder 60 with seizures, 618587
AP3B1	99,2	95,8	100	100	Hermansky-Pudlak syndrome 2, 608233
AP3B2	93,3	89,5	99,8	98,6	Developmental and epileptic encephalopathy 48, 617276
AP3D1	99,8	98,6	100	100	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	99,9	98,7	100	100	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	99,8	98,7	100	100	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,9	98,9	100	100	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,9	71,3	87,9	87,9	Spastic paraplegia 52, autosomal recessive, 614067
APC2	97,6	92,7	99,9	99,1	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APTX	94,9	92,4	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	97	96,6	96,9	96,6	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	100	100	100	100	Periventricular nodular heterotopia 8, 618185
ARFGEF2	99,9	99,1	100	100	Periventricular heterotopia with microcephaly, 608097
ARG1	92,9	92,9	92,9	92,9	Argininemia, 207800
ARHGAP31	99,9	98,8	100	100	Adams-Oliver syndrome 1, 100300
ARHGEF6	99,5	96,2	100	99,9	No OMIM disease ID
ARHGEF9	76,5	74,1	97,2	97,1	Developmental and epileptic encephalopathy 8, 300607
ARID1A	98,1	96,4	100	100	Coffin-Siris syndrome 2, 614607
ARID1B	96,2	95,2	97,9	96,7	Coffin-Siris syndrome 1, 135900
ARID2	99,8	98,5	100	100	Coffin-Siris syndrome 6, 617808
ARL13B	100	99,2	100	100	Joubert syndrome 8, 612291
ARL6	99,9	98,6	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC9	100	99,8	100	100	Joubert syndrome 30, 617622

ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ARSL	99	93	100	99,9	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100	99,9	100	100	Developmental and epileptic encephalopathy 38, 617020
ARX	81	64	91,5	85,7	Lissencephaly, X-linked 2, 300215 Proud syndrome, 300004 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	99,7	98,6	100	100	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASH1L	98,7	98,6	98,7	98,7	Mental retardation, autosomal dominant 52, 617796
ASL	100	99,6	100	100	Argininosuccinic aciduria, 207900
ASNS	99,4	95,2	100	100	Asparagine synthetase deficiency, 615574
ASPA	99,9	98,3	100	100	Canavan disease, 271900
ASPM	99,7	98,2	100	100	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	95,4	87,9	100	100	Citrullinemia, 215700
ASXL1	99,8	99,3	99,8	99,8	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	99,7	98,9	100	100	Shashi-Pena syndrome, 617190
ASXL3	99,9	99,7	100	100	Bainbridge-Ropers syndrome, 615485
ATAD1	99,6	95,1	100	100	Hyperekplexia 4, 618011
ATAD3A	91,9	83,2	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATIC	99,9	99,3	100	100	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100	99,7	100	100	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATN1	99,9	98,2	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	99,9	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	94,1	76,6	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	99,5	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	99,9	98,7	100	100	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1B2	100	99,3	100	100	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP7A	99	96,9	100	100	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	100	99,7	100	100	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATR	99,9	99,4	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	99,4	96,3	100	100	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	100	99,8	100	100	3-methylglutaconic aciduria, type I, 250950
AUTS2	98,2	95,8	100	100	Mental retardation, autosomal dominant 26, 615834
AVPR2	100	99,4	100	100	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
B3GALNT2	93,8	89,4	92,5	92,5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	75,7	69,7	89,8	81,6	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GLCT	99,6	96,3	99,9	99,2	Peters-plus syndrome, 261540
B4GALNT1	99,3	95	100	100	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT7	99,8	97,4	99,9	98,6	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	99,9	99	100	100	No OMIM disease ID
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,8	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989

BBS2	100	99,5	100	100	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,9	99,3	100	100	Bardet-Biedl syndrome 4, 615982
BBS5	99	93,9	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	98,7	95,5	100	100	Bardet-Biedl syndrome 7, 615984
BBS9	92,3	90,4	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCAP31	92,6	83,2	100	99,9	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	99,9	99,2	100	100	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,5	94,4	100	100	Maple syrup urine disease, type Ib, 248600
BCKDK	100	100	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL11A	97,7	96	100	100	Dias-Logan syndrome, 617101
BCL11B	99,1	95,6	98,8	97,3	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCOR	99,6	97,4	100	99,9	Microphthalmia, syndromic 2, 300166
BCORL1	99,6	97,9	100	100	Shukla-Vernon syndrome, 301029
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	99,8	98,3	100	100	Bloom syndrome, 210900
BOLA3	99,4	90,2	100	100	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	96,2	94,3	99,6	98,6	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	91	81,1	100	100	Melanoma, malignant, somatic, 155600 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 114500 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0
BRAT1	99,7	98,2	100	100	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BRF1	99,9	98,4	100	100	Cerebellofaciodental syndrome, 616202
BRPF1	100	100	100	100	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	99,5	97,4	100	100	No OMIM disease ID
BRWD3	99,3	97,2	100	100	Mental retardation, X-linked 93, 300659
BSCL2	100	100	100	100	Neuropathy, distal hereditary motor, type VC, 619112 Lipodystrophy, congenital generalized, type 2, 269700

					Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	83,1	83	83,1	83,1	Biotinidase deficiency, 253260
BUB1B	99,6	98,9	100	100	Colorectal cancer, somatic, 114500 [Premature chromatid separation trait], 176430 Mosaic variegated aneuploidy syndrome 1, 257300
C12orf4	100	99,3	100	100	Mental retardation, autosomal recessive 66, 618221
C12orf57	100	98,9	100	100	Temtamy syndrome, 218340
C12orf65	99,8	98,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C2CD3	95,8	95,6	95,9	95,9	Orofaciodigital syndrome XIV, 615948
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	87,4	85,2	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	99,6	97,3	100	100	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	93,2	90	100	99,9	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Developemental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNA1B	97,5	95,7	99,1	97,7	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	99,9	99,2	100	100	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	98	97,9	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100	99,9	100	100	Developmental and epileptic encephalopathy 69, 618285
CACNA1G	100	99,6	100	100	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA2D2	94	93,2	99,2	97,6	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100	99,2	100	100	Developmental and epileptic encephalopathy 50, 616457
CAMK2A	99,9	99	99,8	98,7	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	100	99,8	100	100	Mental retardation, autosomal dominant 54, 617799
CAMK2G	99,9	98,1	100	100	Mental retardation, autosomal dominant 59, 618522
CAMTA1	100	99,5	100	100	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	100	99,9	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719

CARS2	100	100	100	99,2	Combined oxidative phosphorylation deficiency 27, 616672
CASK	97,3	94,2	100	100	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	99,8	98,3	100	100	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	100	99,3	100	100	Mental retardation, autosomal recessive 3, 608443
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCBE1	99,8	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC115	95,3	90	100	100	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	99,5	97,1	100	100	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	99,6	96,5	100	100	Ritscher-Schinzel syndrome 2, 300963
CCDC32	100	99,5	100	100	Cardiofacioneurodevelopmental syndrome, 619123
CCDC47	99,4	97,5	100	100	Trichohepatoneurodevelopmental syndrome, 618268
CCDC88A	96,4	93,1	97,5	97,5	?PEHO syndrome-like, 617507
CCDC88C	100	99,3	100	100	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCND2	100	100	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	92,6	89,8	100	99,8	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CDC42	97,9	90,9	100	100	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100	99,3	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	99,9	98,7	100	100	Mental retardation, autosomal dominant 3, 612580
CDK10	100	99,9	100	100	Al Kaissi syndrome, 617694
CDK13	98	92,7	100	100	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	100	99,9	100	100	Developmental and epileptic encephalopathy 87, 618916
CDK5RAP2	99,8	98,9	100	100	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	99,7	97,9	100	100	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	91,7	90,2	92,3	91,7	Developmental and epileptic encephalopathy 2, 300672
CDKN1C	88	77,8	99,3	97,3	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDON	100	99,6	100	100	Holoprosencephaly 11, 614226

CENPF	99,8	98,5	100	100	Stromme syndrome, 243605
CENPJ	100	99,6	100	100	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	100	99,2	100	100	Joubert syndrome 25, 616781
CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	99,1	93,6	100	100	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	99,7	98,2	100	100	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	96,1	90	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	99,8	97,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	99,2	93	100	100	Mosaic variegated aneuploidy syndrome 2, 614114
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862
CEP89	96	94,5	100	100	No OMIM disease ID
CERT1	90,2	87,3	100	100	Mental retardation, autosomal dominant 34, 616351
CHAMP1	100	100	100	100	Mental retardation, autosomal dominant 40, 616579
CHD1	99,3	94,9	100	100	Pilarowski-Bjornsson syndrome, 617682
CHD2	99,4	99,2	100	100	Epileptic encephalopathy, childhood-onset, 615369
CHD3	94,8	92,6	99,8	99,5	Snijders Blok-Campeau syndrome, 618205
CHD4	100	99,9	100	100	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	100	99,9	100	100	{Autism, susceptibility to, 18}, 615032
CHKB	100	99,7	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100	99,8	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHRNA4	98,3	96,2	100	100	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CIC	63,3	63,3	100	99,9	Mental retardation, autosomal dominant 45, 617600
CIT	100	99,4	100	100	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	99,7	98,6	100	100	Filippi syndrome, 272440
CLCN4	99,9	98,9	100	100	Raynaud-Claes syndrome, 300114

CLIC2	99,9	96,5	100	100	?Mental retardation, X-linked, syndromic 32, 300886
CLIP1	100	99	100	100	No OMIM disease ID
CLN3	92,5	91,8	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69,3	66,3	72,1	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,9	97,1	100	100	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,5	83,5	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	94,9	94,9	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLTC	100	99,9	100	100	Mental retardation, autosomal dominant 56, 617854
CNKSR2	95,5	90,8	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	99,9	100	100	Holoprosencephaly 12, with or without pancreatic agenesis, 618500 Vissers-Bodmer syndrome, 619033
CNOT2	99,9	99,5	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	99,3	100	100	Developmental and epileptic encephalopathy 60, 617929
CNTNAP2	100	99,8	100	100	{Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA8	81,9	80,7	93,5	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100	100	100	100	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COG1	100	100	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG4	100	99,9	100	100	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	99,7	97,6	100	100	Congenital disorder of glycosylation, type IIi, 613612
COG6	99,1	93,9	100	100	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	100	100	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	99,9	98,6	100	100	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	98,7	97,4	100	100	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519

					?Retinal arteries, tortuosity of, 180000 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A2	100	99,6	100	100	Brain small vessel disease 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,9	89,3	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100	99,5	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	100	97,9	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX15	99,9	98,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX6B1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 7, 619051
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CPLX1	100	100	100	100	Developmental and epileptic encephalopathy 63, 617976
CPS1	100	99,9	100	100	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371
CRADD	99,5	96,3	100	100	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	88,2	87,7	97	92,9	Mental retardation, autosomal recessive 2, 607417
CREBBP	99,7	98,5	100	100	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRLF1	91	89,8	97,9	95,2	Cold-induced sweating syndrome 1, 272430
CRPPA	98,5	94,8	100	99,4	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CSDE1	99,9	99,5	100	100	No OMIM disease ID
CSF1R	99,9	99,3	100	100	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSNK2A1	81,5	77,7	94	94	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100	100	100	100	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	99,8	98,7	100	100	Joubert syndrome 21, 615636
CSTB	99,6	89,8	100	100	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	93,2	86,9	99,5	98,6	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100	99,3	100	100	Mental retardation, autosomal dominant 21, 615502
CTDP1	88,4	84,3	100	99,4	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100	99,8	100	100	Cortical dysplasia, complex, with other brain malformations 9, 618174

CTNNB1	100	99,9	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	93,5	91,1	97,7	95,5	No OMIM disease ID
CTSA	100	100	100	100	Galactosialidosis, 256540
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	99,5	97,3	100	100	No OMIM disease ID
CTU2	99,7	97,7	100	100	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUL4B	98	90,8	99,9	99,2	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX1	96,4	94,8	99,3	98,4	Global developmental delay with or without impaired intellectual development, 618330
CUX2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 67, 618141
CWC27	99,3	96,5	100	100	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXorf56	99,8	96,7	100	100	?Mental retardation, X-linked 107, 301013
CYB5R3	98,4	98	99,8	98,9	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYFIP2	100	99,4	100	100	Developmental and epileptic encephalopathy 65, 618008
CYP27A1	98,9	96,7	100	100	Cerebrotendinous xanthomatosis, 213700
CYP2U1	94,8	91,5	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
D2HGDH	99,2	97,2	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
DARS1	100	99,3	100	100	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	94,9	94,3	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	99,8	98	100	100	Maple syrup urine disease, type II, 248600
DCAF17	98,9	93,3	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	99,8	99,1	100	100	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	91,3	91,2	100	100	Al-Raqad syndrome, 616459

DCX	100	99,9	100	100	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDC	99,7	96,4	100	100	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	100	99,6	100	100	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	85,2	80,7	100	100	Warsaw breakage syndrome, 613398
DDX3X	81,2	78,9	98	96,1	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX59	100	100	100	100	Orofaciodigital syndrome V, 174300
DDX6	97,7	88,7	100	100	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	97,3	88,8	100	98,7	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEGS1	100	100	100	100	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100	99,4	100	100	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100	99,8	100	100	Epilepsy, familial focal, with variable foci 1, 604364
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	99	95	95,2	95,2	Retinitis pigmentosa 59, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861
DHFR	92,1	78,9	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	100	99,7	93,3	93,2	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	99,9	98,9	100	100	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	100	99,9	100	100	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DIAPH1	99,8	99	99,5	98	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900
DIP2B	100	99,3	100	100	Mental retardation, FRA12A type, 136630
DIS3L2	100	99,8	100	100	Perlman syndrome, 267000
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
DLD	100	99,7	100	100	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	99,1	93,2	100	100	Mental retardation, X-linked 90, 300850
DLG4	99,1	99	98,8	98,8	Intellectual developmental disorder 62, 618793
DMD	99,6	98,6	100	100	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DMPK	99,8	98,4	100	100	Myotonic dystrophy 1, 160900

DMXL2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNAJC12	87,4	87,4	100	100	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	98,9	96,2	100	100	3-methylglutaconic aciduria, type V, 610198
DNM1	92,6	89,1	97,4	97,4	Developmental and epileptic encephalopathy 31, 616346
DNMT3A	99,8	98,6	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	100	100	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	99,3	98,9	100	100	Adams-Oliver syndrome 2, 614219
DOCK7	99,8	98,2	100	99,9	Developmental and epileptic encephalopathy 23, 615859
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DONSON	91,7	85,3	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	99,9	98,4	100	100	Coffin-Siris syndrome 7, 618027
DPH1	100	99,9	100	100	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	98,2	91,3	99,7	97,1	Congenital disorder of glycosylation, type Ie, 608799
DPP6	99,7	97,8	99,4	97,6	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPYD	99,7	97,7	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100	99,9	100	100	Dihydropyrimidinuria, 222748
DYM	97,4	96,5	100	100	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	99,9	99,4	100	100	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
DYNC1I2	84,4	68,8	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	99,7	95,8	100	100	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	99,9	99	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277

EDC3	100	99,9	100	100	?Mental retardation, autosomal recessive 50, 616460
EED	96,5	91,4	99,8	98,2	Cohen-Gibson syndrome, 617561
EEF1A2	100	100	99,9	99,1	Developmental and epileptic encephalopathy 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFNB2	100	99,8	100	100	No OMIM disease ID
EFTUD2	100	99,8	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	94,5	93,7	99,6	99,5	Kleefstra syndrome 1, 610253
EIF2AK1	98,8	95,4	100	100	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100	99,7	100	100	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877
EIF2AK3	97,2	94,5	100	100	Wolcott-Rallison syndrome, 226980
EIF2B4	100	99,9	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	100	99,1	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2S3	95,4	89,1	100	100	MEHMO syndrome, 300148
EIF3F	96,8	84,1	100	100	Mental retardation, autosomal recessive 67, 618295
EIF4A3	100	99,5	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	100	99,7	100	100	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELOVL4	100	99,5	100	100	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP2	99,9	98,8	100	100	Mental retardation, autosomal recessive 58, 617270
EMC1	100	99,3	100	100	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EML1	99,7	98,4	100	100	Band heterotopia, 600348
EMX2	100	100	100	100	Schizencephaly, 269160
ENTPD1	100	100	100	100	Spastic paraplegia 64, autosomal recessive, 615683
EP300	99,8	99	100	100	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
EPG5	99,5	98,5	100	100	Vici syndrome, 242840
ERCC1	100	99,3	100	100	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100	99,7	100	100	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	96,9	96,3	100	100	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390

ERCC5	100	99,7	100	100	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100	100	100	100	{Macular degeneration, age-related, susceptibility to, 5}, 613761 {Lung cancer, susceptibility to}, 211980 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	99,5	95,8	100	100	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	100	99,9	100	100	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	98,7	95,2	100	100	Roberts-SC phocomelia syndrome, 268300
ETFB	100	99,8	100	100	Glutaric acidemia IIB, 231680
ETHE1	99,9	97,4	100	100	Ethylmalonic encephalopathy, 602473
EXOSC2	100	100	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	99,5	94,9	100	100	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC9	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 1D, 618065
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100	99,5	100	100	Weaver syndrome, 277590
FA2H	92	83,1	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	100	99,4	100	100	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	99,5	95,4	100	100	Joubert syndrome 36, 618763
FAM20C	100	100	100	99,8	Raine syndrome, 259775
FAR1	97,6	92,8	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	98,8	94,6	100	100	Rajab interstitial lung disease with brain calcifications 1, 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	96,9	92,7	100	100	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	96	93,1	100	99,9	?Mental retardation, autosomal recessive 45, 615979
FDFT1	97,7	96	100	100	Squalene synthase deficiency, 618156

FGD1	97,3	92,8	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	99,9	98,1	100	100	Developmental and epileptic encephalopathy 47, 617166
FGF14	100	100	100	100	Spinocerebellar ataxia 27, 609307
FGFR1	100	99,9	100	100	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,7	97,1	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	99,8	97,7	100	99,8	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	92,1	88,3	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
FKRP	100	100	100	99,9	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	99,7	97	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	99,9	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	98,9	100	100	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100	100	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	85,5	82,5	100	100	Mental retardation, autosomal recessive 47, 616193
FMR1	96,1	92,1	100	100	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624
FOLR1	100	100	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	88,6	82,1	99,2	96,4	Rett syndrome, congenital variant, 613454
FOXJ1	99,9	98,4	100	100	Ciliary dyskinesia, primary, 43, 618699
FOXP1	100	99,8	100	100	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	99,5	99,2	100	100	Speech-language disorder-1, 602081
FOXRED1	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	100	99,4	100	100	Fraser syndrome 1, 219000
FRMD4A	90,7	87,3	96,6	96,6	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819

FRMPD4	97,5	96,5	98,3	98,3	Mental retardation, X-linked 104, 300983
FRRS1L	79,7	69,1	99,2	95,8	Developmental and epileptic encephalopathy 37, 616981
FTCD	95,7	91	100	100	Glutamate formiminotransferase deficiency, 229100
FTO	83,8	83,7	94,2	94,2	{Obesity, susceptibility to, BMIQ14}, 612460 Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	98	93,8	100	100	Mental retardation, X-linked 9/44, 309549
FUCA1	100	99,9	100	100	Fucosidosis, 230000
FUT8	100	99,2	100	100	Congenital disorder of glycosylation with defective fucosylation 1, 618005
GABBR2	96,2	92	99,1	98,4	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, susceptibility to}, 188890 {Nicotine dependence, protection against}, 188890 Developmental and epileptic encephalopathy 59, 617904
GABRA1	100	100	100	100	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 Developmental and epileptic encephalopathy 19, 615744 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRA2	99,7	98,6	100	100	{Alcohol dependence, susceptibility to}, 103780 Developmental and epileptic encephalopathy 78, 618557
GABRA3	98,7	94,4	99,9	98,9	No OMIM disease ID
GABRA5	100	99,9	100	100	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100	100	100	100	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100	99,9	100	100	Developmental and epileptic encephalopathy 92, 617829
GABRB3	99,6	98,2	100	100	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Developmental and epileptic encephalopathy 43, 617113
GABRG2	90,8	90,2	93	93	Febrile seizures, familial, 8, 607681 Developmental and epileptic encephalopathy 74, 618396 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAD1	100	99,9	100	100	?Cerebral palsy, spastic quadriplegic, 1, 603513 Developmental and epileptic encephalopathy 89, 619124
GALC	99,8	98,3	100	100	Krabbe disease, 245200
GALE	100	100	100	100	Galactose epimerase deficiency, 230350
GALNT2	99,6	97	100	100	Congenital disorder of glycosylation, type IIt, 618885
GALT	100	99,7	100	100	Galactosemia, 230400
GAMT	93,1	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	100	100	100	100	GAND syndrome, 615074
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600

GCH1	99,9	95,5	100	100	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCSH	75,7	68,9	100	100	?Glycine encephalopathy, 605899
GDI1	99,8	98,7	100	100	Mental retardation, X-linked 41, 300849
GFAP	91,8	89,7	100	100	Alexander disease, 203450
GFER	99,6	93,9	100	100	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	99,9	99,4	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,9	95,2	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	78,2	58,7	96,9	91,4	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GK	88,9	70,4	100	99,9	Glycerol kinase deficiency, 307030
GLB1	99,9	97,4	100	100	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	89,9	82	100	99,9	Glycine encephalopathy, 605899
GLI2	99,1	97,4	100	99,8	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS3	98,6	98,2	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLUD1	94,2	82,9	100	100	Hyperinsulinism-hyperammonemia syndrome, 606762
GLYCTK	98,8	97,3	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
GNAI1	99	93,9	100	100	No OMIM disease ID
GNAO1	93,8	93,8	100	100	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAS	86,9	85,1	82	81,7	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	Myelodysplastic syndrome, somatic, 614286 Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GNB5	100	98,8	100	100	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	99,7	97,3	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,1	94,3	100	99,9	Mucolipidosis III gamma, 252605
GNS	98,4	94,8	100	100	Mucopolysaccharidosis type IIID, 252940
GOT2	97,5	90,9	100	100	Developmental and epileptic encephalopathy 82, 618721
GPAA1	98,9	95,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	99,1	94,7	100	100	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC4	100	99,6	100	100	Keipert syndrome, 301026
GPHN	100	99,5	100	100	Molybdenum cofactor deficiency C, 615501
GPSM2	99,9	99,2	100	100	Chudley-McCullough syndrome, 604213
GPT2	99,2	93,6	100	99,8	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GRIA3	99,7	96,1	100	99,6	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIA4	99,8	99	100	100	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	96,2	95,4	96,3	96,3	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	99,8	99,2	100	100	Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 Developmental and epileptic encephalopathy 27, 616139
GRIN2D	79,8	65,4	93,9	88,7	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100	99,7	100	100	Fraser syndrome 3, 617667
GRM1	100	99,7	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	99,8	97,7	100	100	No OMIM disease ID
GSS	96,5	96,4	100	100	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	72,5	72,2	72,5	72,5	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100	99,3	100	99,9	Jaberi-Elahi syndrome, 617988
GTPBP3	100	99,8	100	100	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	92,9	91,7	100	100	Mucopolysaccharidosis VII, 253220
H1-4	100	100	100	100	Rahman syndrome, 617537
H4C3	100	100	100	100	No OMIM disease ID
HACE1	100	99,3	100	100	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99	97,5	100	100	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	99,8	97,6	100	100	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	98,3	93,6	100	100	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	98,5	98,2	98,5	98,5	Generalized epilepsy with febrile seizures plus, type 10, 618482 Developmental and epileptic encephalopathy 24, 615871
HDAC4	100	99,8	100	100	No OMIM disease ID

HDAC6	99,5	97,4	100	100	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	86,5	85,1	96,3	94,8	Cornelia de Lange syndrome 5, 300882
HECW2	100	99,1	100	100	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	86	78,9	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	79,9	77,2	100	100	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
HESX1	99,7	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	93,8	93,3	100	100	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	99,6	96,9	100	99,9	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	86,4	86,3	91,2	89,3	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	98,2	88,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIVEP2	100	100	100	100	Mental retardation, autosomal dominant 43, 616977
HK1	100	100	100	100	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HMGCL	100	99,8	100	100	HMG-CoA lyase deficiency, 246450
HNMT	100	99,8	100	100	{Asthma, susceptibility to}, 600807 Mental retardation, autosomal recessive 51, 616739
HNRNPH1	99,4	96,2	100	100	No OMIM disease ID
HNRNPH2	100	100	100	100	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	91,5	82,8	100	100	Au-Kline syndrome, 616580
HNRNPU	99,9	98,9	100	100	Developmental and epileptic encephalopathy 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	99,3	91,8	100	99,3	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HRAS	100	100	100	100	Costello syndrome, 218040 Bladder cancer, somatic, 109800 Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550
HSD17B10	100	99,1	100	100	HSD10 mitochondrial disease, 300438
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPA9	88,5	84,5	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	98,8	93,7	100	100	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	100	99,9	100	100	{Parkinson disease 13}, 610297 3-methylglutaconic aciduria, type VIII, 617248
HUWE1	99,2	95,8	100	100	Mental retardation, X-linked syndromic, Turner type, 309590
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IARS1	100	99,6	100	100	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100	99,9	100	100	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	93,7	90,1	100	100	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
IDS	99,9	98	100	100	Mucopolysaccharidosis II, 309900
IDUA	93,7	86,8	100	100	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IER3IP1	91,9	82,6	100	100	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	99,7	98,4	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT81	93,5	90,1	95	94,9	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	99,5	96,2	100	100	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472

IGF1	100	99,9	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100	99,9	100	100	Insulin-like growth factor I, resistance to, 270450
IKBKG	84,1	77,2	100	100	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291
IL1RAPL1	99,8	98,6	100	100	Mental retardation, X-linked 21/34, 300143
IMPA1	97	87	100	100	Mental retardation, autosomal recessive 59, 617323
INPP5E	97,1	92,7	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
INTS1	99,8	98,5	100	100	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
IQSEC1	89,1	86,3	97,6	95,2	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	96,8	88,6	99,4	98,4	Mental retardation, X-linked 1/78, 309530
IRF2BPL	99,5	95	99,9	99,2	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ISCA2	100	98,8	100	100	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITGA7	99,6	98	100	100	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	100	99,9	100	100	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700
IVD	100	100	100	100	Isovaleric acidemia, 243500
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAM3	100	99,9	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	99,9	99,2	100	100	No OMIM disease ID
KANK1	100	100	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	99,9	99,2	100	100	Koolen-De Vries syndrome, 610443
KAT6A	100	99,8	100	100	Arboleda-Tham syndrome, 616268
KAT6B	99,6	98,3	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KAT8	99,9	98,8	100	100	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100	99,9	100	100	Lissencephaly 6, with microcephaly, 616212
KCNA2	100	99,6	100	100	Developmental and epileptic encephalopathy 32, 616366
KCNA4	100	100	100	100	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284

KCNB1	100	99,6	100	100	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100	100	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	78,6	65,8	95	89,7	Spinocerebellar ataxia 13, 605259
KCNH1	98,7	98,7	98,7	98,7	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	89,3	89	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 mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ6	100	100	100	100	Keppen-Lubinsky syndrome, 614098
KCNK4	99,1	97,4	100	100	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	97,3	97,3	97,3	97,3	Birk-Barel syndrome, 612292
KCNMA1	94,4	93,6	100	100	Liang-Wang syndrome, 618729 {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNQ2	91,3	89,8	100	100	Seizures, benign neonatal, 1, 121200 Developmental and epileptic encephalopathy 7, 613720 Myokymia, 121200
KCNQ3	100	99,4	99,8	99,1	Seizures, benign neonatal, 2, 121201
KCNQ5	97,8	95,5	100	100	Mental retardation, autosomal dominant 46, 617601
KCNT1	96	95,2	98,6	97,3	Epilepsy nocturnal frontal lobe, 5, 615005 Developmental and epileptic encephalopathy 14, 614959
KCNT2	99,4	97,1	100	100	Developmental and epileptic encephalopathy 57, 617771
KCTD3	100	99,7	100	100	No OMIM disease ID
KCTD7	95	95	100	100	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	98,2	95,3	100	100	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	97,5	96,3	100	100	Diets-Jongmans syndrome, 618846
KDM5B	94,6	92,3	93,9	92,9	Mental retardation, autosomal recessive 65, 618109
KDM5C	99,8	97,9	100	100	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	96,1	88,7	100	99,9	Kabuki syndrome 2, 300867
KDM6B	98,8	97,9	100	100	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505

KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1109	99,8	99,2	100	100	Alkuraya-Kucinskas syndrome, 617822
KIDINS220	100	100	100	100	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	97,6	94,8	100	100	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	99,6	97,7	100	100	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	97,4	95,2	98	98	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF2A	99,6	95,6	100	100	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100	99,7	100	100	Retinitis pigmentosa 89, 618955
KIF4A	99,4	95,7	100	100	?Mental retardation, X-linked 100, 300923
KIF5A	100	99,9	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KIF5C	99,9	98,8	99,8	99,8	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1	96,1	96,1	96,1	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL3	99,8	98,9	100	100	No OMIM disease ID
KLF7	100	99,7	100	100	No OMIM disease ID
KLHL15	100	99,7	100	100	Mental retardation, X-linked 103, 300982
KMT2A	100	99,9	99,9	99,4	Wiedemann-Steiner syndrome, 605130
KMT2B	95,8	94	98,7	97,9	Dystonia 28, childhood-onset, 617284
KMT2C	92,2	91	100	100	Kleefstra syndrome 2, 617768
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
KMT2E	99,8	98,5	100	100	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	99,9	99,1	100	100	Mental retardation, autosomal dominant 51, 617788
KNL1	99,2	98,1	98,9	98,8	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100	100	100	100	Mental retardation, autosomal recessive 41, 615637
KRAS	99,5	96,9	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	99,9	99,1	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	97,2	100	100	L-2-hydroxyglutaric aciduria, 236792
LAMA1	100	99,7	100	100	Poretti-Boltshauser syndrome, 615960
LAMA2	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMB1	100	99,9	100	100	Lissencephaly 5, 615191
LAMC3	98,6	97,1	100	99,6	Cortical malformations, occipital, 614115
LAMP2	99,2	95,6	100	100	Danon disease, 300257
LARGE1	100	99,6	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	88,5	78,4	100	100	Alazami syndrome, 615071
LAS1L	99,7	97,3	100	100	Wilson-Turner syndrome, 309585
LIAS	100	99,1	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
LINGO1	100	100	100	100	Mental retardation, autosomal recessive 64, 618103
LINS1	100	99,1	100	100	Mental retardation, autosomal recessive 27, 614340
LMAN2L	100	99,7	100	100	?Mental retardation, autosomal recessive, 52, 616887
LONP1	100	99,8	100	100	CODAS syndrome, 600373
LRP2	100	99,9	100	100	Donnai-Barrow syndrome, 222448
LRPPRC	99,9	99,1	100	100	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
LZTFL1	99,9	99,2	100	100	Bardet-Biedl syndrome 17, 615994

LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 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	99,7	99,3	100	100	Lissencephaly 9 with complex brainstem malformation, 618325
MAF	83,5	78	88,6	82,2	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAG	100	100	100	100	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	93	87,2	100	100	Schaaf-Yang syndrome, 615547
MAN1B1	100	99,7	100	99,9	Rafiq syndrome, 614202
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,8	86,5	100	100	Mannosidosis, beta, 248510
MAOA	100	99,7	99,8	98,5	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAP1B	99,3	97,7	100	100	Periventricular nodular heterotopia 9, 618918
MAP2K1	99,8	97,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,1	100	100	Cardiofaciocutaneous syndrome 4, 615280
MAPK8IP3	99,4	99	100	100	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPRE2	100	99,3	100	100	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	100	99,9	100	100	3MC syndrome 1, 257920
MAST1	100	99,5	100	100	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAT1A	99,7	97,7	100	100	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MBD5	99,9	99,9	100	100	Mental retardation, autosomal dominant 1, 156200
MBOAT7	100	99,5	100	100	Mental retardation, autosomal recessive 57, 617188
MBTPS2	100	99	100	100	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918
MCCC1	100	99,8	100	100	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,9	98,4	100	100	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	99,8	98,4	100	100	Mucolipidosis IV, 252650
MCPH1	100	99,4	100	100	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	98	97,9	100	100	Developmental and epileptic encephalopathy 51, 617339

MECP2	100	98,7	100	99,9	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 {Autism susceptibility, X-linked 3}, 300496 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	100	98,9	100	100	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	99,8	96,7	100	100	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MED13	100	99,9	100	100	Intellectual developmental disorder 61, 618009
MED13L	100	99,8	100	100	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MED17	96,3	93,5	100	100	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100	99,7	100	100	Mental retardation, autosomal recessive 18, 614249
MED25	100	99,8	100	100	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	99,9	96	100	100	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	99,9	99	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	99,3	98,5	99,9	98,3	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	94,3	89,9	100	100	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	99,7	98,5	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100	99,7	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	98,7	95,1	100	100	Keutel syndrome, 245150
MICU1	98,9	95,2	100	100	Myopathy with extrapyramidal signs, 615673
MID1	99,8	98,7	100	100	Opitz GBBB syndrome, type I, 300000
MID2	99,8	98,7	100	100	?Mental retardation, X-linked 101, 300928
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000

MLC1	100	99	100	100	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	96	90,4	100	98,9	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100	100	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100	99,6	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94,4	83,5	89,7	89,7	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MMUT	99,8	98,3	100	100	Methylmalonic aciduria, mut(0) type, 251000
MN1	100	99,3	100	100	Meningioma, 607174 CEBALID syndrome, 618774
MOCS1	99,2	95,1	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,6	99,5	100	100	Molybdenum cofactor deficiency B, 252160
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	100	100	100	100	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,8	98,8	100	100	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPLKIP	100	99,4	100	100	Trichothiodystrophy 4, nonphotosensitive, 234050
MRAS	100	99,6	100	100	Noonan syndrome 11, 618499
MRPS22	99,8	99,1	100	100	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MSL3	84,5	77,4	96,8	96,6	Basilicata-Akhtar syndrome, 301032
MSMO1	96,3	88,9	100	100	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTFMT	100	99,8	100	100	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	97,3	96	100	100	{Schizophrenia, susceptibility to}, 181500 Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTO1	91,3	90,4	91,6	91,4	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100	99,5	100	100	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTR	100	100	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100	99,6	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cbl E type, 236270

MVK	90,9	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYCN	100	99,9	99,3	96,7	Feingold syndrome 1, 164280
MYH9	100	99,3	100	100	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	99,8	98,9	100	100	Griscelli syndrome, type 1, 214450
MYO9A	99,9	99,1	100	100	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYT1L	87	86,2	90,2	90,2	Mental retardation, autosomal dominant 39, 616521
NAA10	99,7	98,5	99,9	99,9	Ogden syndrome, 300855 Microphthalmia, syndromic 1, 309800
NAA15	95,8	91	96,8	96,7	Mental retardation, autosomal dominant 50, 617787
NACC1	100	99,8	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	92,9	89,9	99,9	99,2	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NALCN	100	99,5	99,8	99,8	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	98,3	97,4	100	100	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAXE	100	99,8	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	92	90,6	100	100	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBN	99,9	98,6	100	100	Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135
NCAPG2	99,9	99,2	100	100	Khan-Khan-Katsanis syndrome, 618460
NDE1	100	100	100	100	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDP	100	99,7	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	100	100	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA1	99,9	99,3	100	100	Mitochondrial complex I deficiency, nuclear type 12, 301020

NDUFA11	100	100	100	99,8	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	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	62,6	61,1	100	99,6	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,7	90,6	91,9	90,7	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100	99,2	100	100	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	98	96,1	100	100	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,9	76,9	100	100	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	72	71,5	100	100	Periventricular nodular heterotopia 7, 617201
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	100	99,5	100	100	Mental retardation, X-linked 98, 300912
NF1	92,6	90,2	100	100	Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Watson syndrome, 193520 Neurofibromatosis, type 1, 162200
NFIA	99,2	98,8	99,2	99,2	Brain malformations with or without urinary tract defects, 613735
NFIB	97,4	96,5	100	100	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100	99,5	99,6	98,7	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFU1	98,8	90,8	100	100	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100	99,8	100	100	Congenital disorder of deglycosylation, 615273
NHS	95,4	93,9	100	99,8	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIPBL	98,9	97	100	100	Cornelia de Lange syndrome 1, 122470
NKAP	99,3	95,2	100	100	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
NKX2-1	98,6	85,6	100	100	{Thyroid cancer, nonmedullary, 1}, 188550 Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978

NLGN2	92,9	88,5	100	100	No OMIM disease ID
NLGN3	99,9	99	100	100	{Autism susceptibility, X-linked 1}, 300425 {Asperger syndrome susceptibility, X-linked 1}, 300494
NLGN4X	99,9	98,9	100	99,9	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NONO	100	98,4	100	100	Mental retardation, X-linked, syndromic 34, 300967
NOVA2	99	94,6	96,8	93,3	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPC1	99,6	98,7	100	100	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100	99,6	100	100	Niemann-pick disease, type C2, 607625
NPHP1	100	99	100	100	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NR2F1	100	100	99,1	95,1	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
NRROS	100	100	100	100	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	97,4	96,9	100	99,8	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	100	99,9	100	100	Sotos syndrome 1, 117550
NSD2	99,9	99,2	100	100	No OMIM disease ID
NSDHL	100	98,7	100	100	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	96	95,1	100	100	Mental retardation, autosomal recessive 5, 611091
NT5C2	98	96,5	100	100	Spastic paraplegia 45, autosomal recessive, 613162
NTNG2	98,5	96,7	99,9	99	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	99,8	98,2	100	100	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	100	99,9	100	100	Obesity, hyperphagia, and developmental delay, 613886 Developmental and epileptic encephalopathy 58, 617830

NUBPL	99,7	98,4	100	100	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP62	100	100	100	100	Striatonigral degeneration, infantile, 271930
NUS1	60	44,5	100	100	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	85,2	76,3	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	100	100	100	100	Pseudo-TORCH syndrome 1, 251290
OCRL	99,9	98,6	100	99,9	Lowe syndrome, 309000 Dent disease 2, 300555
ODC1	100	99,8	100	100	Bachmann-Bupp syndrome, 619075
OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGT	99,9	99	100	100	Mental retardation, X-linked 106, 300997
OPA3	100	99	100	100	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPHN1	99,5	97,6	99,9	98,8	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	100	99,4	100	100	Meier-Gorlin syndrome 1, 224690
OSGEP	100	99,4	100	100	Galloway-Mowat syndrome 3, 617729
OTC	100	100	100	100	Ornithine transcarbamylase deficiency, 311250
OTUD6B	99,9	98,8	100	100	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTX2	100	99,7	100	100	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OXR1	99,4	97	100	100	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P4HTM	99	97,4	100	99,4	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PACS1	98,8	96,9	100	100	Schuurs-Hoeijmakers syndrome, 615009
PACS2	99,3	96,2	100	99,8	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	94,1	87,1	100	100	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	100	100	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK1	100	99,6	100	100	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	99,3	95,9	100	99,8	Mental retardation, X-linked 30/47, 300558
PANK2	100	99,3	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200

PANX1	100	100	100	100	Oocyte maturation defect 7, 618550
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX1	92,4	87,9	100	99,6	Otofaciocervical syndrome 2, 615560
PAX6	100	100	100	100	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430
PAX7	100	100	100	100	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX8	100	99,8	100	100	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	100	99,4	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	99,8	97,3	100	100	Pyruvate carboxylase deficiency, 266150
PCCA	99,5	96,7	100	100	Propionicacidemia, 606054
PCCB	97,9	96	98,7	96,2	Propionicacidemia, 606054
PCDH12	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH19	100	98,9	100	100	Developmental and epileptic encephalopathy 9, 300088
PCGF2	100	99,5	100	100	Turnpenny-Fry syndrome, 618371
PCLO	99,7	98,7	100	100	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	99,6	97,1	100	100	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT2	99,8	97,1	100	98,8	Spastic paraplegia 82, autosomal recessive, 618770
PDE4D	95,7	93,5	100	99,8	Acrodysostosis 2, with or without hormone resistance, 614613
PDGFRB	99,2	97,5	100	100	Myeloproliferative disorder with eosinophilia, 131440 Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Premature aging syndrome, Penttinen type, 601812 Myofibromatosis, infantile, 1, 228550
PDHA1	99,4	97,1	100	100	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	99,9	99,4	100	100	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100	100	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94,7	87,6	97,3	96,6	Coenzyme Q10 deficiency, primary, 2, 614651

PDSS2	99,8	97,1	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	100	98,8	100	100	Prolidase deficiency, 170100
PET100	100	99,6	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	100	99,6	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	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,9	98,5	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	99,3	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGAP1	99	94,4	100	100	Mental retardation, autosomal recessive 42, 615802
PGAP2	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	63,5	59,6	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	92,8	79,3	100	100	Phosphoglycerate kinase 1 deficiency, 300653
PGM3	100	99,8	91,7	91,7	Immunodeficiency 23, 615816
PHACTR1	100	99,7	100	100	Developmental and epileptic encephalopathy 70, 618298

PHF21A	100	99,9	100	100	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF6	97,8	88,3	99,9	98,9	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	99,7	96,8	100	100	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	98,6	96,1	100	99,7	Chung-Jansen syndrome, 617991
PI4KA	92,6	88,8	99,9	99,9	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	99,5	96,2	100	100	Joubert syndrome 33, 617767
PIGA	93,8	86,7	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	99,9	97,8	100	100	Developmental and epileptic encephalopathy 80, 618580
PIGC	99,2	90,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100	99,7	100	100	Mental retardation, autosomal recessive 53, 616917
PIGH	82,1	68,1	75,2	74,4	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	99,2	95,1	100	100	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100	100	100	100	CHIME syndrome, 280000
PIGN	93,8	91,5	98,8	98,8	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,8	87,3	100	100	Developmental and epileptic encephalopathy 55, 617599
PIGS	100	100	100	100	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	98,1	98,1	100	100	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100	99,1	100	100	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100	99,8	100	100	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	98	97,8	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	90,7	89,6	99,3	96,1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PITRM1	98,4	96,1	100	100	No OMIM disease ID
PLA2G6	92,2	90,7	92,3	92,3	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLAA	100	99,2	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	100	99,8	100	100	Developmental and epileptic encephalopathy 12, 613722
PLK4	99,9	98,2	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	100	99,2	100	100	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	98,2	90,1	100	100	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLXND1	98,9	96,2	99,7	99,4	No OMIM disease ID
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	97,7	94,2	100	100	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100	99,7	100	100	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	100	100	100	100	Ataxia-oculomotor apraxia 4, 616267 ?Charcot-Marie-Tooth disease, type 2B2, 605589 Microcephaly, seizures, and developmental delay, 613402
PNP	99,8	98,9	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POGZ	99,4	99	100	100	White-Sutton syndrome, 616364
POLA1	99,3	95,4	100	99,9	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLG	100	99,3	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	90,5	87	82,8	82,8	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	99,7	100	100	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,9	98,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	100	99,9	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	99,3	97,5	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	99,4	96,4	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	99,1	100	100	Focal dermal hypoplasia, 305600
POU1F1	100	99,2	100	100	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	73,2	59,6	94,9	83,8	Snijders Blok-Fisher syndrome, 618604
PPM1D	100	99,9	100	100	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	99,9	99,3	100	100	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	97,7	95,3	100	100	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	100	99,6	100	100	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	99,6	96	100	100	No OMIM disease ID
PPP2CA	100	100	100	100	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	91,6	91,5	93,6	93,6	Mental retardation, autosomal dominant 36, 616362
PPP2R5B	100	100	100	100	No OMIM disease ID
PPP2R5C	97,7	93,1	100	100	No OMIM disease ID
PPP2R5D	100	100	100	100	Mental retardation, autosomal dominant 35, 616355
PPP3CA	99,8	98,4	100	100	Developmental and epileptic encephalopathy 91, 617711 Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265
PPT1	90,3	90,3	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100	100	100	100	Renpenning syndrome, 309500

PRKAR1A	99,3	93,5	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	99,9	100	100	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	85	80,6	100	100	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	86,4	86,4	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	98,7	97,2	100	100	No OMIM disease ID
PRSS12	100	99,9	100	100	Mental retardation, autosomal recessive 1, 249500
PRUNE1	93,6	93,5	93,6	93,6	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	95,3	81,6	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMD12	98,6	92,9	100	100	Stankiewicz-Isidor syndrome, 617516
PSPH	100	100	100	100	Phosphoserine phosphatase deficiency, 614023
PTCH1	99,2	97,6	99,9	99,8	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCHD1	100	99,9	100	100	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	100	100	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	99,5	97	100	100	Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 {Meningioma}, 607174
PTF1A	95,8	85,6	98,6	93,3	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935

PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN23	100	100	100	100	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100	100	100	100	No OMIM disease ID
PTS	99,9	99,1	100	100	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100	99,3	100	100	Verheij syndrome, 615583
PUM1	100	99,9	100	100	Spinocerebellar ataxia 47, 617931
PURA	99	95,2	100	99,8	Mental retardation, autosomal dominant 31, 616158
PUS1	100	99,5	99,6	97,2	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100	100	100	100	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100	99,8	100	100	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PYCR1	99,9	97,7	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,1	100	100	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100	100	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100	99,7	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100	99,9	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	99,5	97,4	100	100	Warburg micro syndrome 3, 614222
RAB23	100	99,5	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	99,4	98,9	99,4	99,4	Warburg micro syndrome 1, 600118
RAB3GAP2	99,5	97	100	100	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RAC1	99,9	96,2	100	99,9	Mental retardation, autosomal dominant 48, 617751
RAC3	97,3	94,4	99,7	98,2	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	99,2	96,6	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	94,6	87,9	100	100	No OMIM disease ID
RALGAPA1	74,5	63,9	100	100	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RARB	100	100	100	100	Microphthalmia, syndromic 12, 615524
RARS1	94,2	91,6	94,4	94,3	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100	99,8	100	100	Pontocerebellar hypoplasia, type 6, 611523
RBBP8	100	99,7	100	100	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBFOX1	89,2	88,8	99,2	97,7	No OMIM disease ID
RBM10	99,5	97,1	100	100	TARP syndrome, 311900
RBM28	100	100	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	98,4	92,8	100	100	Adams-Oliver syndrome 3, 614814
RCBTB1	99,9	99,5	100	100	Retinal dystrophy with or without extraocular anomalies, 617175
RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400
RELN	100	99,8	100	100	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320
RERE	96,1	91,3	99,9	99,9	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	97,6	97,2	97,6	97,6	No OMIM disease ID
RFT1	99,8	99,6	100	100	Congenital disorder of glycosylation, type In, 612015
RFX3	100	100	100	100	No OMIM disease ID
RHEB	88,8	75,4	100	100	No OMIM disease ID
RHOBTB2	100	100	100	100	Developmental and epileptic encephalopathy 64, 618004
RIC1	100	99,9	100	100	CATIFA syndrome, 618761
RIMS2	96,7	95,3	97,8	97,7	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RLIM	100	99	100	100	Tonne-Kalscheuer syndrome, 300978
RMND1	100	98,6	100	100	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460

RNASEH2A	100	100	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	80,6	78,1	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	99,5	100	100	Aicardi-Goutieres syndrome 3, 610329
RNASET2	97,4	93,1	100	100	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100	100	100	100	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	99,9	99,2	100	100	Tenorio syndrome, 616260
RNF13	95,2	81,6	100	100	Developmental and epileptic encephalopathy 73, 618379
ROGDI	98,4	95,2	99,9	99,1	Kohlschutter-Tonz syndrome, 226750
ROR2	100	99,9	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	96,7	90,2	100	100	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPIA	98,6	94,9	100	100	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	97,4	89,1	100	100	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPS19	100	99,6	100	100	Diamond-Blackfan anemia 1, 105650
RPS6KA3	98,4	94,5	100	98,9	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RRM2B	100	99,7	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	99,8	96,8	100	100	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	99,5	96,8	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	99,9	98,7	100	100	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	98,9	98	100	100	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,4	97,5	100	100	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100	100	100	100	Mental retardation, autosomal recessive 61, 617773
RXYLT1	99,5	96,8	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SALL1	99,9	99	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480

SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SAMHD1	98,7	98,4	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS1	100	99,3	100	100	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SATB2	99,7	97,4	100	100	Glass syndrome, 612313
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SC5D	100	99,5	100	100	Lathosterolosis, 607330
SCAMP5	100	100	100	100	No OMIM disease ID
SCAPER	99,7	98,2	100	100	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCN1A	99,9	99,5	100	100	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403
SCN1B	98	96,4	99,8	99,3	Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	99,6	97,6	100	100	Episodic ataxia, type 9, 618924 Developmental and epileptic encephalopathy 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	99,8	99,2	100	100	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN8A	100	99,8	100	100	Seizures, benign familial infantile, 5, 617080 Developmental and epileptic encephalopathy 13, 614558 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364
SCO1	97,1	93,8	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCYL1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	100	99,9	100	100	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615

SDHA	85,8	80,4	100	100	Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165 Mitochondrial complex II deficiency, nuclear type 1, 252011
SEC31A	99,3	97,1	100	100	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651
SEMA3E	99,2	98,9	100	100	?CHARGE syndrome, 214800
SEPSECS	100	100	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,9	99,5	100	100	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	98,3	90,5	98,9	97	Mental retardation, autosomal dominant 58, 618106
SETBP1	99,9	98,7	100	100	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	100	99,8	100	100	Epilepsy, early-onset, with or without developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	98,2	97,5	100	100	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100	99,9	100	100	Luscan-Lumish syndrome, 616831
SETD5	100	99,8	98	98	Mental retardation, autosomal dominant 23, 615761
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SGSH	94,4	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK2	97,7	97,6	98,9	98,9	{Autism susceptibility 17}, 613436
SHANK3	91,6	81,5	96	91,9	{Schizophrenia 15}, 613950 Phelan-McDermid syndrome, 606232
SHH	100	99,5	100	100	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	99,9	99,4	100	100	Noonan syndrome-like with loose anagen hair 1, 607721
SHROOM4	100	99	100	100	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIAH1	100	100	100	100	No OMIM disease ID
SIK1	98,7	94,4	100	100	Developmental and epileptic encephalopathy 30, 616341
SIL1	99,2	96,7	100	100	Marinesco-Sjogren syndrome, 248800
SIN3A	100	99	100	100	Witteveen-Kolk syndrome, 613406
SIX3	99,9	98,6	100	100	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	99,3	94,9	100	99,4	Shprintzen-Goldberg syndrome, 182212
SLC12A5	83,9	83,8	97,4	97,4	Developmental and epileptic encephalopathy 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	100	100	100	100	Agenesis of the corpus callosum with peripheral neuropathy, 218000

SLC13A5	100	99,9	100	100	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A2	99,2	93,7	100	100	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	99,6	97	100	100	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC19A3	97,8	97,6	98,7	98,7	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	99,9	99,6	100	100	{?Schizophrenia susceptibility 18}, 615232 Dicarboxylic aminoaciduria, 222730
SLC1A2	96,1	95,4	100	100	Developmental and epileptic encephalopathy 41, 617105
SLC1A4	99	95,8	100	100	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	95,8	88,6	99,5	97,8	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	99,9	99,5	100	100	Developmental and epileptic encephalopathy 39, 612949
SLC25A15	99,8	98,1	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	98,6	95,8	100	100	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99,4	99,3	99,8	99,8	Fontaine progeroid syndrome, 612289
SLC2A1	92,8	92,8	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 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC33A1	99,9	98,9	100	100	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	100	99,7	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	99,9	98,4	100	100	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	80,7	78,6	81,1	81	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	99,9	98,7	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC39A14	100	99,4	93,5	93,5	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC4A4	99,8	99,2	100	100	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A6	100	100	100	100	Neurodegeneration, infantile-onset, biotin-responsive, 618973
SLC6A1	96,7	96,7	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A17	100	100	100	100	Mental retardation, autosomal recessive 48, 616269

SLC6A19	100	100	100	100	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	100	100	100	100	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135
SLC6A8	93,5	81,6	100	99,8	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100	100	100	100	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC9A6	95,2	91,6	100	98,4	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	97,6	90,3	99,9	99,5	Intellectual developmental disorder, X-linked 108, 301024
SMAD4	100	99,9	100	100	Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350
SMARCA1	99,6	97,5	100	99,6	No OMIM disease ID
SMARCA2	96,7	96,2	97,4	96,8	Nicolaides-Baraitser syndrome, 601358
SMARCA4	99,9	99	100	100	{Rhabdoid tumor predisposition syndrome 2}, 613325 Coffin-Siris syndrome 4, 614609
SMARCB1	100	100	100	100	Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091 Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCC2	99	96,6	100	100	Coffin-Siris syndrome 8, 618362
SMARCD1	94,2	89,3	100	100	Coffin-Siris syndrome 11, 618779
SMARCE1	95,6	88,8	100	100	{Meningioma, familial, susceptibility to}, 607174 Coffin-Siris syndrome 5, 616938
SMC1A	100	98,7	100	99,8	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	95,2	91	100	100	Cornelia de Lange syndrome 3, 610759
SMG9	100	100	100	100	Heart and brain malformation syndrome, 616920
SMOC1	99,9	98,4	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	99,4	94,2	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	91,5	78,5	100	99,9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	100	99,9	100	100	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	100	100	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528

SNIP1	98,9	97,1	100	100	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRPB	100	99,3	100	100	Cerebrocostomandibular syndrome, 117650
SNRPN	100	97	100	100	Prader-Willi syndrome, 176270
SNX14	99,6	95,9	100	100	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100	99,5	100	100	No OMIM disease ID
SOBP	97,5	92,9	97	95,3	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	98,8	94,9	100	100	ZTTK syndrome, 617140
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100	99,2	100	100	Noonan syndrome 9, 616559
SOX10	99,9	97,9	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	91,4	75,2	100	99,5	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX4	97,3	90,9	99,9	98,2	Coffin-Siris syndrome 10, 618506
SOX5	99,9	98,9	100	100	Lamb-Shaffer syndrome, 616803
SOX6	99,9	99,4	100	100	Tolchin-Le Caignec syndrome, 618971
SPART	99,7	96,8	100	100	Troyer syndrome, 275900
SPAST	99,8	98,7	100	100	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	100	99,7	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPECC1L	96	95,7	97,8	96,2	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPG11	100	99,3	100	100	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPOCK1	100	99,5	100	100	No OMIM disease ID
SPOP	100	100	100	100	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829
SPR	99,8	96,3	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100	98,9	100	100	Legius syndrome, 611431
SPTAN1	99,1	98,6	100	100	Developmental and epileptic encephalopathy 5, 613477

SPTBN2	100	99,3	99,9	99,9	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
SPTBN4	97,3	91	100	100	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SRCAP	99,4	98,9	100	100	Floating-Harbor syndrome, 136140
SRD5A3	99,9	99,1	100	100	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SRPX2	99,8	96,5	100	100	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SSR4	100	99,7	100	100	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	68,8	68,6	95,3	95,2	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	85	84,2	98,7	98,4	Salt and pepper developmental regression syndrome, 609056
STAG1	99,6	97,3	100	100	Mental retardation, autosomal dominant 47, 617635
STAG2	97,6	89,4	99,9	98,7	Mullegama-Klein-Martinez syndrome, 301022 Holoprosencephaly 13, X-linked, 301043
STAMBP	100	99,4	100	100	Microcephaly-capillary malformation syndrome, 614261
STIL	100	99,8	100	100	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	100	99,8	100	100	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100	98,9	100	100	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STT3A	100	100	100	100	Congenital disorder of glycosylation, type Iw, 615596
STT3B	100	99,6	100	100	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	100	100	100	100	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	96,8	96,5	100	100	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	89,5	82,2	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	99,9	99,8	100	100	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUMF1	97,5	90,8	100	100	Multiple sulfatase deficiency, 272200
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
SUPT16H	98,6	93,6	100	100	No OMIM disease ID
SURF1	89,4	88,2	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	91,4	86,2	100	100	Imagawa-Matsumoto syndrome, 618786
SVBP	100	100	100	100	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYN1	81,9	73,2	100	99,6	?Intellectual developmental disorder, X-linked 50, 300115 Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	97,8	87,2	100	100	No OMIM disease ID

SYNGAP1	99,4	98,1	100	100	Mental retardation, autosomal dominant 5, 612621
SYNJ1	99,9	99,4	100	100	Developmental and epileptic encephalopathy 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	99,9	96,7	100	100	Mental retardation, X-linked 96, 300802
SYT1	99,8	98,5	100	100	Baker-Gordon syndrome, 618218
SZT2	99,6	99,5	100	99,9	Developmental and epileptic encephalopathy 18, 615476
TAF1	99,8	97,7	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	99,9	98,6	100	100	Mental retardation, autosomal recessive 40, 615599
TAF6	99,8	98,9	100	100	Alazami-Yuan syndrome, 617126
TANC2	100	99,5	100	100	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	99,5	97,9	100	100	No OMIM disease ID
TASP1	99,7	98,8	100	100	Suleiman-El-Hattab syndrome, 618950
TAT	100	100	100	100	Tyrosinemia, type II, 276600
TBC1D20	94,2	94,2	100	99,9	Warburg micro syndrome 4, 615663
TBC1D23	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100	100	100	100	Developmental and epileptic encephalopathy 16, 615338 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal recessive 86, 614617
TBC1D2B	99,3	97,6	98,5	97,8	No OMIM disease ID
TBC1D7	100	99,3	100	100	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	96,2	94,4	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,8	97,5	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	99,1	96,8	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	96,5	84,9	100	100	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
TBP	100	99,9	100	100	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136

TBR1	99,9	97,9	100	100	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TCF20	100	100	100	100	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF4	100	99,8	100	100	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	99,9	98,8	100	100	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TCTN2	100	99,5	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	99,4	100	100	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100	100	100	100	Spastic paraplegia 49, autosomal recessive, 615031
TECR	100	99	100	100	Mental retardation, autosomal recessive 14, 614020
TELO2	99,7	96,2	100	100	You-Hoover-Fong syndrome, 616954
TENM3	100	99,7	100	100	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TET3	94,4	94,4	100	100	Beck-Fahrner syndrome, 618798
TFAP2A	99,4	94,3	100	100	Branchiooculofacial syndrome, 113620
TGDS	99,4	96,8	100	100	Catel-Manzke syndrome, 616145
TGFBR1	93,7	93,6	99	96,3	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGIF1	100	100	100	100	Holoprosencephaly 4, 142946
TH	99,3	96,1	100	100	Segawa syndrome, recessive, 605407
THOC2	98,8	93,7	100	100	Mental retardation, X-linked 12/35, 300957
THOC6	100	100	100	100	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100	99,7	100	100	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIMM50	98,3	94,4	100	100	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	98,1	90,6	100	100	Mohr-Tranebjaerg syndrome, 304700
TINF2	100	100	100	100	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TKFC	100	99,5	100	100	Triokinase and FMN cyclase deficiency syndrome, 618805

TKT	98,7	97,8	98,7	98,7	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	99,1	95,1	100	100	Mental retardation, autosomal dominant 57, 618050
TMCO1	88	87,4	88	88	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM106B	99,9	98,8	100	100	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100	99,9	100	100	Joubert syndrome 14, 614424
TMEM240	100	100	100	100	Spinocerebellar ataxia 21, 607454
TMEM63A	100	99,9	100	100	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TMEM70	98	93,9	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	99,5	97,1	100	99,9	{Autism, susceptibility to, X-linked 6}, 300872
TMTC3	99,6	96,5	100	100	Lissencephaly 8, 617255
TMX2	100	99,8	100	100	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNIK	100	99,3	100	100	Mental retardation, autosomal recessive 54, 617028
TNRC6B	100	99,8	100	100	No OMIM disease ID
TOE1	100	100	100	100	Pontocerebellar hypoplasia, type 7, 614969
TP53RK	92,5	79,6	100	100	Galloway-Mowat syndrome 4, 617730
TPI1	99,8	97,5	100	100	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	99,9	98,2	100	100	Thyroid dyshormonogenesis 2A, 274500
TPP1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TPRKB	81,1	75,9	81,9	81,9	Galloway-Mowat syndrome 5, 617731
TRAF7	100	99,8	100	100	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100	100	100	100	Seckel syndrome 9, 616777
TRAPPC11	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC4	100	100	100	100	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	99,9	98	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100	99,6	100	100	Mental retardation, autosomal recessive 13, 613192

TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 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	99,3	97,2	100	100	No OMIM disease ID
TRIO	99,2	97,5	99,3	98,4	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIP12	99,9	99,2	100	100	Mental retardation, autosomal dominant 49, 617752
TRIT1	100	100	100	100	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	99,4	96,2	100	100	Mental retardation, autosomal recessive 68, 618302
TRMT10A	100	99,7	100	100	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	99,5	96,5	100	100	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM3	100	99,5	100	100	No OMIM disease ID
TRRAP	99,9	99,5	100	100	Developmental delay with or without dysmorphic facies and autism, 618454 ?Deafness, autosomal dominant 75, 618778
TSC1	99,8	98,8	100	100	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100	99,6	100	100	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	79	77,2	100	100	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100	99,6	100	100	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	96,3	94,3	99,9	98,9	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	100	99,5	94,9	94,9	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	81,5	73,8	100	99,2	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	100	99,3	100	100	Trichohepatoenteric syndrome 1, 222470
TTC5	100	99,9	100	100	No OMIM disease ID
TTC8	99,6	98,1	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464

TTI2	100	100	100	100	Mental retardation, autosomal recessive 39, 615541
TUBA1A	99,9	97	100	100	Lissencephaly 3, 611603
TUBA8	99,9	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	97,3	93,9	99,8	99,8	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB2A	97	95,7	100	100	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	98,3	96,9	100	100	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	95,9	94	97,1	96	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	99,7	96,2	97	97	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	99,2	96,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100	99,3	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	100	99,5	100	100	Mental retardation, autosomal recessive 7, 611093
TWIST1	100	98,9	97,2	92,3	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
U2AF2	99,9	98,3	100	100	No OMIM disease ID
UBA5	97,8	86,8	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE2A	99,7	96	100	99,7	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	99,1	94,8	100	100	Angelman syndrome, 105830
UBE3B	100	99,9	100	100	Kaufman oculocerebrofacial syndrome, 244450
UBR1	99,9	99,1	98	98	Johanson-Blizzard syndrome, 243800
UBTF	100	99,4	100	100	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	100	100	100	100	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	74	69,4	100	100	Leukodystrophy, hypomyelinating, 14, 617899
UGDH	99,9	99,1	100	100	Developmental and epileptic encephalopathy 84, 618792
UGP2	99	98,6	96,3	96,3	Developmental and epileptic encephalopathy 83, 618744
UNC13A	99,3	97,7	100	100	No OMIM disease ID
UNC80	97,9	97,4	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801

UPB1	100	100	100	100	Beta-ureidopropionase deficiency, 613161
UPF3B	92,2	84,1	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	91,3	87,9	94,8	94,8	Hao-Fountain syndrome, 616863
USP9X	98,2	92,9	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	99,5	97,7	100	100	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VARS1	100	99,9	100	100	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VLDLR	100	99,8	100	100	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	94,9	93,6	100	100	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	99,5	98,2	99,5	99,4	Cohen syndrome, 216550
VPS37A	91,3	78,2	100	100	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	91,5	90,7	100	99,3	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,7	98,5	100	100	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	100	99,7	100	100	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	100	99,7	100	100	Desanto-Shinawi syndrome, 616708
WARS2	100	99,4	100	100	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	99,9	96,5	100	100	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	99,1	95,5	100	100	?Mental retardation, autosomal recessive 43, 615817
WDFY3	100	99,6	100	100	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	98,2	94,4	98,1	98,1	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR13	99,9	98,6	100	100	No OMIM disease ID
WDR26	88,7	83,9	94,2	91,7	Skraban-Deardorff syndrome, 617616
WDR37	86,5	86,2	86,5	86,5	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100	100	100	100	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45	98,1	92,4	100	100	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	98	89,2	100	100	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	100	99,5	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	99,9	100	100	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WWOX	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 12, 614322 Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211
XPA	99,6	95,6	100	100	Xeroderma pigmentosum, group A, 278700
XRCC4	99,9	99,3	100	100	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,4	89,6	98,1	94,8	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777
YME1L1	99	95,2	100	100	?Optic atrophy 11, 617302
YWHAE	100	100	100	100	No OMIM disease ID
YWHAG	100	100	100	100	Developmental and epileptic encephalopathy 56, 617665
YY1	100	99,8	100	100	Gabriele-de Vries syndrome, 617557
ZBTB11	99,9	99,6	100	100	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100	99,9	100	100	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZBTB18	100	99,9	100	99,8	Mental retardation, autosomal dominant 22, 612337
ZBTB20	100	100	100	100	Primrose syndrome, 259050
ZBTB24	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	99,9	98,9	100	100	Mental retardation, autosomal recessive 56, 617125
ZC4H2	100	99	100	100	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZDHHC9	99,9	93,8	100	100	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	99,9	99,1	97,4	97,4	Mowat-Wilson syndrome, 235730
ZFYVE26	100	99,1	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	98,7	98,5	95,7	Holoprosencephaly 5, 609637
ZMIZ1	99,4	98,4	100	100	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYND11	100	99,6	100	100	Mental retardation, autosomal dominant 30, 616083
ZNF142	100	99,9	100	100	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	99,9	99,6	100	100	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260

ZNF292	99,6	98,3	99,6	99,6	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100	99,9	100	100	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	99,9	99,3	100	100	No OMIM disease ID
ZNF41	100	99,6	100	100	No OMIM disease ID
ZNF462	100	99,9	100	100	Weiss-Kruszka syndrome, 618619
ZNF711	99,8	98,2	100	100	Mental retardation, X-linked 97, 300803
ZSWIM6	95,5	91,9	94,9	92,1	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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors