INTELLECTUAL DISABILITY GENE PANEL DG 2.16 (1252 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AAAS	102,3	100.0%	99.6%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS	103,7	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AASS	131,5	99.9%	99.4%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	83,2	99.9%	98.3%	GABA-transaminase deficiency, 613163
ABCC8	125,8	100.0%	99.9%	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	142,6	100.0%	99.7%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	87,4	77.2%	75.0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	129	99.8%	98.4%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	180,9	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACAD9	124,3	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	148,2	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACAT1	110,2	99.7%	98.3%	Alpha-methylacetoacetic aciduria, 203750
ACO2	115,3	95.8%	89.5%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	123,7	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	145,8	99.9%	99.1%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	102,8	98.6%	93.5%	Mental retardation, X-linked 63, 300387
ACTB	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310

ACTG1	116,3	100.0%	100.0%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTL6A	127,7	99.9%	98.8%	No OMIM phenotype
ACVR1	136,9	100.0%	99.9%	Fibrodysplasia ossificans progressiva, 135100
ACY1	118,5	100.0%	98.6%	Aminoacylase 1 deficiency, 609924
ADAM22	133,9	99.9%	99.4%	?Epileptic encephalopathy, early infantile, 61, 617933
ADAR	109,2	99.9%	99.3%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	128	100.0%	99.9%	Mental retardation, autosomal recessive 36, 615286
ADGRG1	147,2	100.0%	100.0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADK	102,4	99.8%	98.0%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	190,5	100.0%	100.0%	Helsmoortel-van der Aa syndrome, 615873
ADSL	138,6	99.2%	98.6%	Adenylosuccinase deficiency, 103050
AFF2	107,8	99.8%	98.8%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	99,8	99.9%	99.0%	CHOPS syndrome, 616368
AFG3L2	98,3	95.9%	86.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGO2	120,4	99.1%	99.0%	No OMIM phenotype {Epithelial ovarian cancer,reduced risk,association with} (Permuth-Wey (2011) Cancer Res 71,3896)
AHCY	111,6	99.9%	97.7%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	148,5	99.7%	98.3%	Xia-Gibbs syndrome, 615829
AHI1	129,8	99.7%	98.3%	Joubert syndrome 3, 608629
AIFM1	90	99.8%	96.7%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	79,4	99.1%	92.4%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	117,6	96.1%	89.3%	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	82,3	99.2%	94.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937

ALDH18A1	113,7	100.0%	99.8%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH3A2	113,5	95.3%	94.3%	Sjogren-Larsson syndrome, 270200
ALDH4A1	123,9	100.0%	99.8%	Hyperprolinemia, type II, 239510
ALDH5A1	91	99.3%	93.2%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	66,7	93.5%	86.1%	Epilepsy, pyridoxine-dependent, 266100
ALG1	46,5	53.2%	50.2%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	129,3	96.8%	96.3%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	155,7	100.0%	99.9%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	77,3	98.5%	92.1%	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG2	103,2	100.0%	100.0%	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ALG6	101,6	99.1%	95.6%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118,5	96.6%	96.2%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	113	100.0%	99.6%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
ALX4	157,1	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMER1	98,2	99.8%	98.9%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	97,4	99.8%	98.9%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	132,3	100.0%	99.9%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	142,7	100.0%	100.0%	Glycine encephalopathy, 605899
ANK3	139,9	99.4%	99.0%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	111,6	100.0%	99.9%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000

ANKLE2	144,7	100.0%	99.8%	?Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	119,6	99.2%	97.1%	KBG syndrome, 148050
ANO10	106	98.9%	96.3%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	108,3	99.0%	96.9%	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S1	101	100.0%	99.8%	MEDNIK syndrome, 609313
AP1S2	55	75.3%	68.6%	Mental retardation, X-linked syndromic 5, 304340
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	125,6	99.4%	97.6%	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	125,2	98.4%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	121	99.9%	98.4%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	106,6	99.8%	98.8%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	129,3	99.7%	98.1%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	66,2	78.5%	71.3%	Spastic paraplegia 52, autosomal recessive, 614067
APC2	122,4	99.9%	98.7%	?Sotos syndrome 3, 617169
APOPT1	NC	NC	NC	Mitochondrial complex IV deficiency, 220110
APTX	96,3	94.1%	91.3%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	141,9	96.7%	96.6%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARFGEF2	125,2	99.7%	98.7%	Periventricular heterotopia with microcephaly, 608097
ARG1	159,1	100.0%	100.0%	Argininemia, 207800
ARHGAP31	141,4	99.8%	98.7%	Adams-Oliver syndrome 1, 100300
ARHGEF6	115,1	98.7%	94.4%	Mental retardation, X-linked 46, 300436
ARHGEF9	51,5	76.1%	71.3%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	134,4	99.4%	98.4%	Coffin-Siris syndrome 2, 614607
ARID1B	139,6	99.5%	99.2%	Coffin-Siris syndrome 1, 135900
ARID2	156,4	99.8%	98.5%	Coffin-Siris syndrome 6, 617808
ARL13B	102,2	100.0%	99.4%	Joubert syndrome 8, 612291
ARL6	100,3	99.8%	98.2%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC9	124,8	100.0%	99.3%	Joubert syndrome 30, 617622

ARSA	138,5	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSE	80,5	97.9%	89.2%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	108,9	100.0%	99.2%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	49,3	87.3%	79.2%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	125,7	99.3%	97.2%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASH1L	143,7	98.7%	98.5%	Mental retardation, autosomal dominant 52, 617796
ASL	123,6	100.0%	98.5%	Argininosuccinic aciduria, 207900
ASNS	82,8	98.6%	92.2%	Asparagine synthetase deficiency, 615574
ASPA	116,1	99.7%	96.9%	Canavan disease, 271900
ASPM	111,6	99.7%	98.0%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	97,4	95.0%	87.1%	Citrullinemia, 215700
ASXL1	132,4	100.0%	99.5%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	140,9	99.7%	98.8%	Shashi-Pena syndrome, 617190
ASXL3	138,1	99.7%	99.1%	Bainbridge-Ropers syndrome, 615485
ATAD1	65,2	99.3%	89.8%	Hyperekplexia 4, 618011
ATAD3A	90,3	93.6%	87.5%	Harel-Yoon syndrome, 617183
ATIC	113,9	100.0%	99.7%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	134,7	99.9%	99.0%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATN1	155,1	99.9%	99.1%	Dentatorubro-pallidoluysian atrophy, 125370
ATP1A1	111,1	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	161,7	100.0%	99.5%	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481

ATP1A3	159,8	100.0%	100.0%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A2	143,2	100.0%	99.8%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP2	44,9	88.4%	64.2%	?Parkinsonism with spasticity, X-linked, 300911 Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	117,4	99.9%	99.0%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V1A	133,4	99.6%	97.3%	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1B2	120,3	100.0%	99.1%	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455
ATP7A	111,2	99.5%	96.7%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	115,2	99.9%	99.5%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATR	144,6	99.8%	98.6%	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
ATRX	89,2	99.1%	95.5%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	127	100.0%	99.7%	3-methylglutaconic aciduria, type I, 250950
AUTS2	130,6	99.5%	97.7%	Mental retardation, autosomal dominant 26, 615834
AVPR2	133,1	100.0%	99.8%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B3GALNT2	93,9	92.9%	91.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	81,7	82.6%	77.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GLCT	96,6	99.7%	99.1%	Peters-plus syndrome, 261540

B4GALNT1	151,2	99.8%	97.9%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT7	123,9	99.8%	98.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	136,9	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BBS1	146,4	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	158,1	100.0%	99.9%	Bardet-Biedl syndrome 10, 615987
BBS12	187,1	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	150,7	99.9%	99.6%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	110,2	99.9%	99.2%	Bardet-Biedl syndrome 4, 615982
BBS5	98,5	98.0%	93.3%	Bardet-Biedl syndrome 5, 615983
BBS7	142,9	99.1%	96.5%	Bardet-Biedl syndrome 7, 615984
BBS9	113,6	98.6%	94.4%	Bardet-Biedl syndrome 9, 615986
BCAP31	73,4	93.2%	78.3%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	176,9	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	123,3	98.6%	92.8%	Maple syrup urine disease, type Ib, 248600
BCKDK	203,5	100.0%	100.0%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL11A	145,5	99.5%	98.0%	Dias-Logan syndrome, 617101
BCL11B	127,4	99.9%	98.0%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCOR	102,7	98.8%	95.3%	Microphthalmia, syndromic 2, 300166
BCS1L	147,9	100.0%	100.0%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	111	99.6%	98.0%	Bloom syndrome, 210900
BOLA3	48,1	99.9%	92.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	143,6	96.3%	94.6%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755

BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRAT1	142	100.0%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRF1	109	99.8%	98.1%	Cerebellofaciodental syndrome, 616202
BRPF1	161,5	100.0%	100.0%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	122,1	99.9%	98.5%	No OMIM phenotype
BRWD3	106,4	99.2%	95.6%	Mental retardation, X-linked 93, 300659
BSCL2	105,2	100.0%	100.0%	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BTD	126,6	99.9%	99.7%	Biotinidase deficiency, 253260
BUB1B	122	99.8%	98.7%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf4	128,2	99.9%	99.3%	Mental retardation, autosomal recessive 66, 618221
C12orf57	145,6	100.0%	100.0%	Temtamy syndrome, 218340
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C2CD3	116,9	95.8%	95.2%	Orofaciodigital syndrome XIV, 615948
C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	93,2	99.6%	95.7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751

CA8	107,5	99.7%	97.6%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	92,4	97.8%	94.7%	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1C	141	99.9%	99.1%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	127,4	98.0%	97.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	120,9	99.8%	99.2%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1G	148,6	100.0%	99.8%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA2D1	94,2	99.0%	94.5%	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077) Histiocytoid cardiomyopathy (Cataldo (2014) Cardiol Young epub) West syndrome (Hino-Fukuyo (2015) Hum Genet 134,
CAD	136,7	99.9%	99.2%	Epileptic encephalopathy, early infantile, 50, 616457
CAMK2A	117,3	100.0%	99.8%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	111,8	100.0%	100.0%	Mental retardation, autosomal dominant 54, 617799
CAMTA1	179,5	100.0%	99.7%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	144,9	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CARS2	128,2	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	85,1	99.5%	94.5%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CBL	126	97.3%	97.0%	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563

CBS	123,3	99.9%	99.0%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	135	100.0%	99.5%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	111,7	99.0%	97.1%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	75,3	99.8%	99.1%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC115	77,9	88.9%	87.1%	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	120,5	99.7%	97.5%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	97,5	98.8%	94.5%	Ritscher-Schinzel syndrome 2, 300963
CCDC88A	92,8	99.3%	96.7%	?PEHO syndrome-like, 617507
CCDC88C	108,2	100.0%	99.4%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCND2	135,7	100.0%	100.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	86,8	90.6%	87.0%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CDC42	90,2	97.6%	89.1%	Takenouchi-Kosaki syndrome, 616737
CDC6	139,4	99.9%	99.8%	?Meier-Gorlin syndrome 5, 613805
CDH11	126,5	100.0%	100.0%	Elsahy-Waters syndrome, 211380
CDH15	157,7	100.0%	99.8%	Mental retardation, autosomal dominant 3, 612580
CDK10	131,3	100.0%	100.0%	Al Kaissi syndrome, 617694
CDK13	126,5	99.9%	98.2%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK5RAP2	107	99.8%	98.8%	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	140,6	99.6%	96.7%	No OMIM phenotype
CDKL5	100	95.1%	93.1%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1C	100,1	89.8%	81.7%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDON	107	100.0%	99.0%	Holoprosencephaly 11, 614226
CENPF	139,9	99.8%	98.7%	Stromme syndrome, 243605
CENPJ	136	99.9%	99.2%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	104	99.3%	97.5%	Joubert syndrome 25, 616781

CEP120	131,7	100.0%	99.4%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	89,9	98.8%	92.6%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	144,4	99.7%	98.2%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	82,6	97.3%	91.7%	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CEP57	85,1	98.7%	91.6%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP83	108,8	99.4%	96.6%	Nephronophthisis 18, 615862
CEP89	127,5	97.4%	94.7%	No OMIM phenotype Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138) ?Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679)
CHAMP1	172,9	100.0%	100.0%	Mental retardation, autosomal dominant 40, 616579
CHD1	108,1	98.2%	91.9%	Pilarowski-Bjornsson syndrome, 617682
CHD2	123,9	99.3%	99.0%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	95	97.7%	94.0%	Snijders Blok-Campeau syndrome, 618205
CHD4	111,8	100.0%	99.9%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	128,4	100.0%	99.8%	{Autism, susceptibility to, 18}, 615032
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	123,2	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
CHRNA4	109,8	99.9%	99.2%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CIC	72,7	64.7%	63.3%	Mental retardation, autosomal dominant 45, 617600
CIT	101,4	99.9%	98.5%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	156	99.9%	98.9%	Filippi syndrome, 272440
CLCN4	105,7	99.9%	98.9%	Raynaud-Claes syndrome, 300114
CLIC2	73,7	99.9%	96.8%	?Mental retardation, X-linked, syndromic 32, 300886

CLIP1	118,1	99.9%	98.8%	No OMIM phenotype Intellectual disability, autosomal recessive (Larti (2015) Eur J Hum Genet 23,331)
CLN3	114,7	92.6%	91.9%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	138,7	99.9%	98.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	132,3	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	144,5	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	135,7	100.0%	100.0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLTC	153,1	100.0%	99.7%	Mental retardation, autosomal dominant 56, 617854
CNKSR2	89	98.5%	92.3%	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	199,8	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNOT1	127,2	100.0%	99.7%	No OMIM phenotype
CNOT2	132,5	99.9%	99.4%	No OMIM phenotype
CNOT3	145,1	100.0%	99.9%	No OMIM phenotype
CNPY3	78,4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 60, 617929
CNTNAP2	127,1	100.0%	99.8%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COASY	172,7	100.0%	100.0%	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COG1	108,4	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209
COG4	94,5	100.0%	99.6%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	126,3	99.9%	98.4%	Congenital disorder of glycosylation, type IIi, 613612
COG6	90,4	99.1%	96.0%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	106,1	100.0%	99.7%	Congenital disorder of glycosylation, type IIe, 608779
COG8	145	100.0%	98.5%	Congenital disorder of glycosylation, type IIh, 611182

COL4A1	95,7	99.6%	97.3%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	109	100.0%	99.1%	Brain small vessel disease 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	133,1	99.7%	97.8%	Mental retardation, autosomal dominant 34, 616351
COLEC11	180,6	100.0%	100.0%	3MC syndrome 2, 265050
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	220,4	100.0%	99.9%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	87,7	99.9%	98.3%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	139,1	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
CPLX1	102,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 63, 617976
CPS1	133,8	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CRADD	113,2	100.0%	98.2%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	125,2	88.1%	87.3%	Mental retardation, autosomal recessive 2, 607417
CREBBP	110,7	99.4%	97.0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRLF1	126,4	93.2%	90.7%	Cold-induced sweating syndrome 1, 272430
CSNK2A1	104,5	93.7%	89.2%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	129,9	100.0%	100.0%	No OMIM phenotype
CSPP1	119	100.0%	99.1%	Joubert syndrome 21, 615636

CSTB	70	99.3%	90.9%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	101,2	94.3%	86.7%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTCF	128,8	99.9%	98.9%	Mental retardation, autosomal dominant 21, 615502
CTDP1	128,1	95.1%	88.0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	106	100.0%	99.6%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNB1	127,4	100.0%	99.9%	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNND2	99,5	96.6%	92.5%	No OMIM phenotype Autism (Turner (2015) Nature 520,51) Intellectual disability (Hofmeister (2015) J Med Genet 52,111)
CTSA	132,9	100.0%	99.9%	Galactosialidosis, 256540
CTSD	171	99.8%	97.8%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	114,2	99.6%	97.5%	No OMIM phenotype ?Autism (Iossifov (2012) Neuron 74,285)
CUL4B	78	97.6%	89.3%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX1	117	97.5%	95.1%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	120,9	99.9%	99.3%	Epileptic encephalopathy, early infantile, 67, 618141
CWC27	84,5	99.8%	97.5%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	103,2	99.9%	99.3%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXorf56	73,4	99.1%	92.7%	?Mental retardation, X-linked 107, 301013
CYB5R3	152,1	99.2%	98.3%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP27A1	173	100.0%	99.7%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	134,3	98.4%	95.5%	Spastic paraplegia 56, autosomal recessive, 615030

D2HGDH	142	100.0%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAG1	189	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	125,4	99.9%	99.0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	126,8	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	109,9	99.6%	96.9%	Maple syrup urine disease, type II, 248600
DCAF17	90,4	99.9%	97.9%	Woodhouse-Sakati syndrome, 241080
DCC	118,6	100.0%	99.8%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCHS1	149,4	100.0%	99.9%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	128	100.0%	99.6%	Al-Raqad syndrome, 616459
DCX	90,5	99.9%	98.4%	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067
DDC	97,9	99.5%	95.0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	129,6	100.0%	99.5%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	100,7	86.7%	81.2%	Warsaw breakage syndrome, 613398
DDX3X	73,5	86.2%	82.9%	Mental retardation, X-linked 102, 300958
DDX59	141,5	100.0%	99.8%	Orofaciodigital syndrome V, 174300
DEAF1	113,4	99.8%	97.6%	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DENND5A	99,2	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	124,5	99.9%	99.7%	Epilepsy, familial focal, with variable foci 1, 604364
DHCR24	155,8	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861

DHFR	50	94.1%	83.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	122,4	99.9%	98.8%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	161,5	100.0%	100.0%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DIAPH1	101,7	99.9%	99.6%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	128	100.0%	99.9%	Mental retardation, FRA12A type, 136630
DIS3L2	143,3	100.0%	99.8%	Perlman syndrome, 267000
DKC1	91,2	99.8%	97.7%	Dyskeratosis congenita, X-linked, 305000
DLD	119,2	99.9%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	79,3	99.2%	92.1%	Mental retardation, X-linked 90, 300850
DLG4	147,6	100.0%	99.9%	no OMIM phenotype Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394)
DMD	108,2	99.4%	98.0%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	151,3	100.0%	99.9%	Myotonic dystrophy 1, 160900
DNAJC12	140,7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	93,8	98.4%	92.3%	3-methylglutaconic aciduria, type V, 610198
DNM1	139,8	94.7%	92.3%	Epileptic encephalopathy, early infantile, 31, 616346
DNMT3A	122,9	99.7%	98.2%	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	116,4	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK6	121,5	99.6%	98.6%	Adams-Oliver syndrome 2, 614219
DOCK7	120,7	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DOLK	157,2	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DONSON	90,2	99.0%	92.4%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	87,5	100.0%	99.9%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	99,8	99.6%	96.7%	Coffin-Siris syndrome 7, 618027

DPH1	161,6	100.0%	100.0%	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	134,7	95.2%	88.2%	Congenital disorder of glycosylation, type Ie, 608799
DPP6	122,4	99.9%	98.6%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPYD	141,6	99.5%	96.4%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	117,5	100.0%	99.8%	Dihydropyrimidinuria, 222748
DYM	103,3	97.4%	95.5%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	140,6	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	130,4	100.0%	99.9%	Mental retardation, autosomal dominant 7, 614104
EBF3	140,9	100.0%	99.8%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	63,2	99.5%	95.2%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	103,8	100.0%	99.7%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDC3	113,7	100.0%	99.0%	?Mental retardation, autosomal recessive 50, 616460
EED	85,3	99.5%	96.3%	Cohen-Gibson syndrome, 617561
EEF1A2	188,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFNB2	147,8	100.0%	99.5%	No OMIM phenotype
EFTUD2	103,2	100.0%	99.2%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	127,7	94.6%	94.2%	Kleefstra syndrome 1, 610253
EIF2AK3	134,2	99.5%	96.3%	Wolcott-Rallison syndrome, 226980
EIF2S3	81,4	96.7%	88.4%	MEHMO syndrome, 300148
EIF3F	63,1	98.7%	88.5%	Mental retardation, autosomal recessive 67, 618295
EIF4A3	87,5	100.0%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	109,4	99.9%	99.0%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731

ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELP2	120,6	99.8%	98.0%	Mental retardation, autosomal recessive 58, 617270
EMC1	105,7	100.0%	98.9%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EML1	129,9	100.0%	99.6%	Band heterotopia, 600348
EMX2	155,2	100.0%	100.0%	Schizencephaly, 269160
ENTPD1	125	100.0%	99.9%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	165,5	99.7%	98.6%	Colorectal cancer, somatic, 114500 Menke-Hennekam syndrome 2, 618333 Rubinstein-Taybi syndrome 2, 613684
EPG5	110,3	99.3%	97.9%	Vici syndrome, 242840
ERCC1	85,7	100.0%	98.1%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	128	100.0%	99.8%	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	92	99.9%	98.4%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC5	126,3	99.9%	99.5%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	158,2	100.0%	99.9%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC8	82,8	98.9%	90.0%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	115,3	100.0%	99.2%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	115,6	99.4%	97.3%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	116,8	100.0%	100.0%	Glutaric acidemia IIB, 231680

ETHE1	97,3	99.9%	97.8%	Ethylmalonic encephalopathy, 602473
EXOSC2	110,3	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	125,1	96.4%	87.8%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC9	132,5	99.6%	95.1%	Pontocerebellar hypoplasia, type 1D, 618065
EXTL3	184,1	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	130	99.4%	97.6%	Weaver syndrome, 277590
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	125,4	100.0%	99.4%	Leukodystrophy, hypomyelinating, 5, 610532
FAM20C	145,2	100.0%	100.0%	Raine syndrome, 259775
FAR1	73,7	97.2%	91.8%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARSB	78	97.4%	92.7%	Rajab interstitial lung disease with brain calcifications, 613658
FAT4	190,3	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBXL3	188,9	100.0%	100.0%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	168,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	86	98.2%	93.7%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	111,5	99.6%	97.3%	?Mental retardation, autosomal recessive 45, 615979
FGD1	86,7	98.4%	93.0%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	100,4	100.0%	99.9%	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	214,2	100.0%	100.0%	Spinocerebellar ataxia 27, 609307
FGFR1	122,6	100.0%	99.6%	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440

FGFR2	113,1	97.7%	96.8%	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	138,5	100.0%	99.6%	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	124,7	100.0%	99.8%	Thauvin-Robinet-Faivre syndrome, 617107
FIGN	131,2	100.0%	100.0%	No OMIM phenotype
FKRP	153,3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155

FKTN	107,5	99.7%	96.1%	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNA	142,7	100.0%	99.9%	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLVCR1	146,1	99.9%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	124,8	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	109,6	86.6%	84.2%	Mental retardation, autosomal recessive 47, 616193
FMR1	78,8	96.3%	91.0%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FOLR1	107,4	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	142	97.8%	88.7%	Rett syndrome, congenital variant, 613454
FOXP1	114,1	100.0%	99.8%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	129,2	99.5%	98.7%	Speech-language disorder-1, 602081
FOXRED1	121	99.8%	98.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	119,2	99.9%	99.2%	Fraser syndrome 1, 219000
FREM2	152,4	100.0%	99.5%	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666
FRMD4A	115,2	91.4%	90.5%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819

FRMPD4	108,4	99.6%	97.2%	Mental retardation, X-linked 104, 300983
FRRS1L	99,1	85.5%	79.1%	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	116,8	98.7%	95.2%	Glutamate formiminotransferase deficiency, 229100
FTO	95,8	83.8%	83.6%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	122,9	99.2%	95.1%	Mental retardation, X-linked 9/44, 309549
FUCA1	125,9	100.0%	99.9%	Fucosidosis, 230000
FUT8	130,4	99.9%	99.2%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
GABBR2	109,2	98.2%	94.6%	Epileptic encephalopathy, early infantile, 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, protection against}, 188890 {Nicotine dependence, susceptibility to}, 188890
GABRA1	164,2	100.0%	99.8%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRA3	82,2	98.9%	95.1%	No OMIM phenotype
GABRB1	169	99.9%	99.8%	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	129,2	100.0%	99.9%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	131,7	99.5%	97.8%	Epileptic encephalopathy, early infantile, 43, 617113 {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	126,4	91.1%	89.7%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAD1	112,7	99.9%	99.7%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	102,9	99.8%	98.8%	Krabbe disease, 245200
GALE	140	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALT	152,6	100.0%	100.0%	Galactosemia, 230400
GAMT	112,5	98.3%	91.5%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	97,4	100.0%	99.1%	Mental retardation, autosomal dominant 18, 615074
GATM	137,3	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GCDH	145,9	100.0%	99.1%	Glutaricaciduria, type I, 231670

GCH1	84,8	100.0%	99.5%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	32,1	88.4%	69.8%	?Glycine encephalopathy, 605899
GDI1	136,4	99.5%	97.9%	Mental retardation, X-linked 41, 300849
GFAP	103,7	91.9%	91.4%	Alexander disease, 203450
GFM1	104,2	99.9%	99.0%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	121,3	98.9%	95.6%	Combined oxidative phosphorylation deficiency 39, 618397
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB1	150,9	100.0%	99.9%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	45,3	92.6%	75.4%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	44,1	84.9%	63.9%	Glycerol kinase deficiency, 307030
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	59,2	90.6%	79.2%	Glycine encephalopathy, 605899
GLI2	158,2	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	139,5	100.0%	99.3%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GLIS3	123,7	100.0%	99.5%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLUD1	65,5	98.1%	87.5%	Hyperinsulinism-hyperammonemia syndrome, 606762

GLYCTK	161,3	100.0%	99.5%	D-glyceric aciduria, 220120
GM2A	122	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMPPA	147,2	100.0%	99.8%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	211,8	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAO1	152,8	93.8%	93.8%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAS	211,3	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNB1	145,2	100.0%	100.0%	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB5	113	99.9%	97.5%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	127,2	99.5%	96.8%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	148	100.0%	99.3%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	177,6	100.0%	98.5%	Mucolipidosis III gamma, 252605
GNS	94,5	99.6%	95.2%	Mucopolysaccharidosis type IIID, 252940
GPAA1	123,1	100.0%	98.3%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	75,7	98.7%	92.7%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	144,7	99.9%	98.8%	Molybdenum cofactor deficiency C, 615501
GPSM2	119,6	99.8%	99.0%	Chudley-McCullough syndrome, 604213

GPT2	121,7	100.0%	99.4%	Mental retardation, autosomal recessive 49, 616281
GRIA3	82,4	98.6%	92.1%	Mental retardation, X-linked 94, 300699
GRIA4	124	99.7%	98.1%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	146,8	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	121,7	96.2%	95.4%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	166,1	100.0%	99.9%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2A	131,2	100.0%	100.0%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	158	99.8%	99.0%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	82,8	91.9%	79.5%	Epileptic encephalopathy, early infantile, 46, 617162
GRIP1	111,1	100.0%	99.3%	Fraser syndrome 3, 617667
GRM1	156,6	100.0%	99.9%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRN	174,1	100.0%	100.0%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GSE1	121,3	100.0%	99.7%	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237)
GSS	93,3	100.0%	99.2%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	81,8	99.9%	95.9%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	128,9	99.7%	98.6%	Jaberi-Elahi syndrome, 617988
GTPBP3	164,7	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	99,5	92.5%	90.5%	Mucopolysaccharidosis VII, 253220
HACE1	136,2	99.9%	99.1%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	111,1	99.3%	98.8%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975

HADHA	72,9	96.3%	89.3%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HAX1	137,4	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	92,4	99.2%	95.2%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	104,4	99.3%	95.8%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	137,5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	119,6	100.0%	99.9%	No OMIM phenotype Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706) Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219) ?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HDAC6	113,9	99.7%	97.7%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	108,1	100.0%	99.2%	Cornelia de Lange syndrome 5, 300882
HECW2	109,3	99.9%	98.7%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	118,3	95.5%	88.7%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	142,1	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	95,1	80.0%	76.1%	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HESX1	66,2	100.0%	98.7%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	106,3	93.7%	92.4%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	163	99.7%	98.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800

HGSNAT	98,3	87.2%	86.2%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	69,9	96.3%	79.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIST1H1E	112,1	100.0%	100.0%	Rahman syndrome, 617537
HIST1H4C	99,7	100.0%	100.0%	No OMIM phenotype
HIVEP2	162,8	100.0%	100.0%	Mental retardation, autosomal dominant 43, 616977
HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMGCL	119,4	99.9%	98.7%	HMG-CoA lyase deficiency, 246450
HNMT	133,1	100.0%	99.9%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPH2	126,2	100.0%	100.0%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	61,1	88.1%	79.9%	Au-Kline syndrome, 616580
HNRNPU	149,9	99.9%	99.3%	Epileptic encephalopathy, early infantile, 54, 617391
HOXA1	164	100.0%	100.0%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HPD	148,1	100.0%	99.7%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	59,8	98.3%	88.2%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HRAS	182,3	100.0%	100.0%	Bladder cancer, somatic, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Thyroid carcinoma, follicular, somatic, 188470
HSD17B10	92,4	100.0%	98.4%	HSD10 mitochondrial disease, 300438
HSD17B4	109,4	96.3%	93.6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPA9	82,6	89.5%	84.2%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	74,3	98.1%	92.5%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	132,6	100.0%	99.6%	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297

HUWE1	79,3	99.1%	94.3%	Mental retardation, X-linked syndromic, Turner type, 300706
HYLS1	156,6	100.0%	100.0%	Hydrolethalus syndrome, 236680
IARS	125,4	99.9%	99.0%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	142,9	100.0%	99.9%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IDS	100,9	99.9%	97.1%	Mucopolysaccharidosis II, 309900
IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	106,3	94.3%	82.8%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	110,8	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT81	92,3	93.6%	89.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	99,1	98.8%	93.2%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	98	100.0%	99.8%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	114,9	100.0%	99.6%	Insulin-like growth factor I, resistance to, 270450
IKBKG	60,1	88.1%	78.8%	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL1RAPL1	99,7	99.9%	98.1%	Mental retardation, X-linked 21/34, 300143
IMPA1	72,1	96.7%	86.7%	Mental retardation, autosomal recessive 59, 617323
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	88,8	100.0%	99.3%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404

IQSEC2	73,1	95.8%	87.9%	Mental retardation, X-linked 1/78, 309530
IRF2BPL	176,5	99.0%	96.7%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ISCA2	105,1	99.8%	95.8%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISPD	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGA7	129,2	99.7%	98.2%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	130,2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	131,2	100.0%	99.7%	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	100	100.0%	99.9%	Isovaleric acidemia, 243500
JAG1	133,7	99.2%	97.1%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAM3	126,9	100.0%	99.9%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	135,3	99.9%	99.3%	No OMIM phenotype ?Rett syndrome (Saez (2016) Genet Med 18,378) ?Congenital heart disease in 22q11.2 deletion syndrome patients (Guo (2015) Am J Hum Genet 97,869) ?Autism spectrum disorder (Saez (2016) Genet Med 18,378) ?Intellectual disabilit
KALRN	124,4	99.9%	99.3%	{Coronary heart disease, susceptibility to, 5}, 608901
KANK1	119,2	100.0%	99.8%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	139,1	99.8%	98.6%	Koolen-De Vries syndrome, 610443
KAT6A	148,4	100.0%	99.7%	Mental retardation, autosomal dominant 32, 616268
KAT6B	155,7	99.9%	99.1%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	154,2	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNA2	126,3	100.0%	99.7%	Epileptic encephalopathy, early infantile, 32, 616366

KCNA4	123	100.0%	100.0%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNB1	129,7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	170,9	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	112,7	90.4%	72.9%	Spinocerebellar ataxia 13, 605259
KCNH1	148,4	98.7%	98.3%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	148,6	89.2%	88.1%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	199,7	100.0%	100.0%	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ6	157,2	100.0%	99.9%	Keppen-Lubinsky syndrome, 614098
KCNK9	171,2	100.0%	100.0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	118,3	91.5%	90.2%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	110,4	99.9%	98.7%	Seizures, benign neonatal, 2, 121201
KCNQ5	135	99.4%	97.7%	Mental retardation, autosomal dominant 46, 617601
KCNT1	131,2	96.0%	95.1%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	154,9	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	130,9	100.0%	98.8%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	121,6	99.0%	97.0%	Mental retardation, autosomal recessive 65, 618109
KDM5C	102,8	99.5%	97.0%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	97,7	95.3%	87.8%	Kabuki syndrome 2, 300867
KIAA0586	117,7	97.0%	93.0%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1109	140,8	99.9%	98.9%	Alkuraya-Kucinskas syndrome, 617822

KIDINS220	137,5	100.0%	99.8%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	92,1	97.8%	94.5%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	116,6	99.6%	97.9%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	115	99.7%	97.6%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1BP	161,1	96.1%	96.0%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF2A	105,7	99.6%	96.8%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	77,4	98.1%	91.5%	?Mental retardation, X-linked 100, 300923
KIF5C	109,9	99.9%	99.0%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	127,5	99.9%	99.1%	Mental retardation, autosomal dominant 4, 612581
KLF7	126	100.0%	99.3%	No OMIM phenotype
KLHL15	140,4	100.0%	99.7%	Mental retardation, X-linked 103, 300982
KMT2A	133	100.0%	99.9%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2B	141,1	96.9%	93.5%	Dystonia 28, childhood-onset, 617284
KMT2C	138,9	91.8%	90.4%	Kleefstra syndrome 2, 617768
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
KMT5B	165,5	100.0%	99.6%	Mental retardation, autosomal dominant 51, 617788
KNL1	105,5	99.0%	97.2%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	145,7	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637

KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
L1CAM	126,6	99.9%	98.6%	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	124	99.0%	96.7%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	116	100.0%	99.5%	Poretti-Boltshauser syndrome, 615960
LAMA2	130,6	100.0%	99.5%	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMB1	142,9	100.0%	99.7%	Lissencephaly 5, 615191
LAMC3	148,1	99.7%	98.6%	Cortical malformations, occipital, 614115
LAMP2	92,3	97.9%	92.8%	Danon disease, 300257
LARGE1	115,2	100.0%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	75,5	88.8%	75.2%	Alazami syndrome, 615071
LAS1L	78,6	99.6%	95.7%	Wilson-Turner syndrome, 309585
LGI4	99,9	99.4%	96.7%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LIAS	125,3	99.9%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	173,4	100.0%	99.8%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500

LINGO1	201,9	100.0%	100.0%	Mental retardation, autosomal recessive 64, 618103
LINS1	133,6	100.0%	99.3%	Mental retardation, autosomal recessive 27, 614340
LMAN2L	109,3	100.0%	99.2%	?Mental retardation, autosomal recessive, 52, 616887
LONP1	148	100.0%	100.0%	CODAS syndrome, 600373
LRP2	139,2	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRPPRC	129,3	100.0%	99.6%	Leigh syndrome, French-Canadian type, 220111
LZTFL1	117	99.8%	99.2%	Bardet-Biedl syndrome 17, 615994
LZTR1	143,6	100.0%	99.7%	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MAB21L1	171,6	100.0%	100.0%	No OMIM phenotype
MAB21L2	237,8	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	128,6	99.9%	99.2%	Lissencephaly 9 with complex brainstem malformation, 618325
MAF	88,9	87.2%	83.0%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAG	160,7	100.0%	100.0%	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	120,4	97.9%	93.1%	Schaaf-Yang syndrome, 615547
MAN1B1	125,5	100.0%	99.8%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	128,6	99.9%	98.6%	Mannosidosis, alpha-, types I and II, 248500
MANBA	118,3	99.5%	97.5%	Mannosidosis, beta, 248510
MAOA	98,9	100.0%	99.0%	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAP1B	127,2	99.9%	99.4%	No OMIM phenotype
MAP2K1	92,3	99.5%	96.3%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	124,2	98.5%	94.1%	Cardiofaciocutaneous syndrome 4, 615280
MAPK8IP3	162	100.0%	99.9%	No OMIM phenotype
MAPRE2	158,4	99.7%	98.1%	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	131,1	100.0%	99.3%	3MC syndrome 1, 257920
MAT1A	144,1	99.8%	98.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	147,7	99.9%	99.8%	Mental retardation, autosomal dominant 1, 156200

MBOAT7	110	100.0%	99.7%	Mental retardation, autosomal recessive 57, 617188
MBTPS2	111,2	99.9%	98.6%	?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014
MCCC1	137,6	100.0%	99.4%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	119	100.0%	99.7%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	157,1	99.9%	99.0%	Mucolipidosis IV, 252650
MCPH1	133,1	99.9%	98.5%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	109,4	98.0%	97.9%	Epileptic encephalopathy, early infantile, 51, 617339
MECP2	124,8	100.0%	98.5%	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MECR	108,2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	85,1	99.5%	95.5%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13	148,7	99.9%	99.5%	No OMIM phenotype
MED13L	108,5	99.9%	99.6%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	132,4	97.5%	94.7%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	133,6	99.8%	98.4%	Mental retardation, autosomal recessive 18, 614249
MED25	132,7	100.0%	99.7%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	127,9	99.4%	95.5%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	144	100.0%	99.5%	Carpenter syndrome 2, 614976

MEIS2	123,6	100.0%	99.8%	Cleft palate, cardiac defects, and mental retardation, 600987
METTL23	116,9	100.0%	100.0%	Mental retardation, autosomal recessive 44, 615942
MFF	86,2	93.7%	89.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	114,3	100.0%	99.3%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	121,3	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	145,4	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066
MGP	134,2	98.7%	94.6%	Keutel syndrome, 245150
MICU1	103,3	98.8%	96.5%	Myopathy with extrapyramidal signs, 615673
MID1	124,1	99.8%	97.4%	Opitz GBBB syndrome, type I, 300000
MID2	108,7	99.5%	97.2%	?Mental retardation, X-linked 101, 300928
MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	92,4	99.6%	97.8%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	96,7	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	95,7	99.4%	96.5%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168,5	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	94,6	100.0%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	196	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	81,2	92.7%	79.5%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	91,2	98.8%	95.7%	Molybdenum cofactor deficiency A, 252150
MOCS2	137,7	99.6%	99.5%	Molybdenum cofactor deficiency B, 252160
MOGS	141	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	102,4	100.0%	99.6%	Congenital disorder of glycosylation, type If, 609180
MPDZ	128,3	99.6%	98.2%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219

MPLKIP	104,3	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRPS22	134,6	99.9%	98.7%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MSL3	73,5	94.7%	83.7%	No OMIM phenotype
MSMO1	51,6	95.8%	88.5%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTFMT	132,5	100.0%	99.8%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTOR	112	99.9%	99.1%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTR	131,4	99.9%	99.4%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	131,1	100.0%	99.0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	NC	NC	NC	Methylmalonic aciduria, mut(0) type, 251000
MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYCN	173,8	100.0%	100.0%	Feingold syndrome 1, 164280
MYH9	128,5	99.6%	98.5%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	109	99.7%	98.6%	Griscelli syndrome, type 1, 214450
MYT1L	144,7	100.0%	99.8%	Mental retardation, autosomal dominant 39, 616521
NAA10	105	100.0%	98.8%	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	95,7	97.6%	94.5%	Mental retardation, autosomal dominant 50, 617787

NACC1	169,5	100.0%	100.0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAGA	121,7	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	117,7	97.1%	94.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	117	99.7%	98.7%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	120,7	97.6%	97.2%	Combined oxidative phosphorylation deficiency 24, 616239
NBEA	125,1	91.9%	90.2%	No OMIM phenotype Autism, idiopathic (Castermans (2003) J Med Genet 40, 352) ?Schizophrenia (Fromer (2014) Nature 506, 179) ?Obesity, extreme (Mariman (2015) Physiol Genomics 47,225) ?Tetralogy of Fallot (Silversides (2012) PloS Genet 8)
NBN	93,8	99.8%	98.4%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NDE1	89,3	100.0%	99.6%	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDP	91,6	100.0%	99.5%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	188,4	100.0%	100.0%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	184,9	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	116	99.8%	97.4%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160,8	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	162,6	100.0%	99.6%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFAF3	141	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	124,9	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFS1	143,5	99.9%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,1	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	124,8	90.7%	90.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230

NDUFS4	144,5	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	111,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	140,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	156,8	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	141,7	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	74,2	92.4%	77.3%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	93,7	72.3%	71.5%	Periventricular nodular heterotopia 7, 617201
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEXMIF	132	100.0%	99.5%	Mental retardation, X-linked 98, 300912
NF1	106,2	92.5%	89.4%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFIA	149,5	100.0%	98.8%	Brain malformations with or without urinary tract defects, 613735
NFIX	174,4	100.0%	99.7%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFU1	61,8	97.4%	82.1%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	135,4	100.0%	99.7%	Congenital disorder of deglycosylation, 615273
NHS	111	98.5%	96.0%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIPBL	124,9	98.8%	96.9%	Cornelia de Lange syndrome 1, 122470
NKX2-1	88,8	100.0%	99.7%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NLGN3	124	99.9%	99.0%	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	147,9	99.5%	97.4%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NONO	78,1	99.1%	95.3%	Mental retardation, X-linked, syndromic 34, 300967
NPC1	117,8	100.0%	99.2%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	124,7	100.0%	99.9%	Niemann-pick disease, type C2, 607625

NPHP1	121,2	99.8%	98.5%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NR2F1	222,3	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	140,4	100.0%	100.0%	No OMIM phenotype
NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NRXN1	141,6	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	147	100.0%	99.8%	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSD2	126,8	99.5%	98.0%	No OMIM phenotype
NSDHL	125,8	99.7%	97.1%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	94,7	97.6%	93.5%	Mental retardation, autosomal recessive 5, 611091
NT5C2	121,2	97.9%	96.3%	Spastic paraplegia 45, autosomal recessive, 613162
NTRK1	133	100.0%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	136,2	100.0%	99.9%	Epileptic encephalopathy, early infantile, 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUBPL	102	98.9%	95.5%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP62	111,8	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
NUS1	53,3	71.5%	44.1%	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
OAT	68,2	81.7%	70.1%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	173,9	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OCRL	106,2	99.8%	98.3%	Dent disease 2, 300555 Lowe syndrome, 309000

ODC1	120,8	99.9%	98.5%	{Colonic adenoma recurrence, reduced risk of}, 114500
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGT	106,5	99.9%	98.4%	Mental retardation, X-linked 106, 300997
OPHN1	78,3	98.9%	95.0%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	90,3	99.9%	98.7%	Meier-Gorlin syndrome 1, 224690
OSGEP	98,1	100.0%	97.3%	Galloway-Mowat syndrome 3, 617729
OTC	111,4	100.0%	99.7%	Ornithine transcarbamylase deficiency, 311250
OTUD6B	117,7	99.8%	99.3%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTX2	127,4	100.0%	99.3%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P4HTM	163,1	100.0%	98.9%	No OMIM phenotype
PACS1	106,7	100.0%	99.4%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	155,4	100.0%	99.3%	Epileptic encephalopathy, early infantile, 66, 618067
PAFAH1B1	77	92.0%	82.8%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	126,4	100.0%	100.0%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	85,3	98.6%	93.7%	Mental retardation, X-linked 30/47, 300558
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	131,8	100.0%	100.0%	No OMIM phenotype Intellectual disability, sensorineural hearing loss, skeletal defects and primary ovarian failure (Shao (2016) J Biol Chem 291,12432)
PARN	127,3	99.9%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX1	189,5	97.0%	92.1%	?Otofaciocervical syndrome 2, 615560

PAX6	116,5	100.0%	99.8%	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX8	94,4	100.0%	99.8%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	111,7	99.9%	98.2%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	155,4	99.9%	98.7%	Pyruvate carboxylase deficiency, 266150
PCCA	99,2	99.3%	95.5%	Propionicacidemia, 606054
PCCB	111,8	99.3%	96.9%	Propionicacidemia, 606054
PCDH19	176,6	99.9%	98.9%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	95,1	99.4%	97.0%	Turnpenny-Fry syndrome, 618371
PCLO	142,1	99.8%	99.0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	115,4	99.7%	97.7%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	102,7	95.8%	94.4%	Acrodysostosis 2, with or without hormone resistance, 614613
PDHA1	85,3	98.9%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	129	99.9%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	129,1	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	104,8	96.7%	87.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	117,4	100.0%	99.6%	Prolidase deficiency, 170100
PET100	87,9	98.0%	87.6%	Mitochondrial complex IV deficiency, 220110
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539

PEX10	113,3	99.9%	97.4%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	87,9	100.0%	99.4%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	120,6	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	179,6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	140,8	98.6%	94.8%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	84,9	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	134,9	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	94,3	100.0%	99.6%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	113,9	99.9%	99.2%	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	106,5	98.5%	92.0%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGAP1	110,9	99.1%	95.8%	Mental retardation, autosomal recessive 42, 615802
PGAP2	134,7	100.0%	99.5%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	70,3	63.5%	59.9%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	44,7	90.9%	75.9%	Phosphoglycerate kinase 1 deficiency, 300653
PGM3	149,3	99.9%	99.6%	Immunodeficiency 23, 615816
PHF21A	93,4	100.0%	99.6%	No OMIM phenotype
PHF6	60,3	98.2%	87.9%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	74,4	99.2%	95.8%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815

PHIP	130,8	99.3%	97.2%	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991
PI4KA	91,8	93.7%	89.7%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	85,9	99.3%	92.2%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	143,4	100.0%	99.5%	Mental retardation, autosomal recessive 53, 616917
PIGL	122,1	99.7%	99.6%	CHIME syndrome, 280000
PIGN	106,3	93.6%	91.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	144,5	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	159,3	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	124,4	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	145	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	90,3	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	127,7	100.0%	99.8%	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3R2	104,3	93.9%	90.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953

PLAA	163	99.7%	98.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	134,9	100.0%	99.7%	Epileptic encephalopathy, early infantile, 12, 613722
PLK4	149,7	99.8%	98.2%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	112,8	99.7%	97.7%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	95,3	99.6%	95.3%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLXND1	122,1	99.8%	98.0%	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6) Truncus arteriosus (Ta-Shma (2013) Am J Med Genet A 161,3115) {Diabetic nephropathy,association with} (McKnight (2009) Hugo J 3,77)
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	108,1	99.1%	95.9%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121,6	100.0%	99.2%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	109	100.0%	99.9%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	108,6	100.0%	99.5%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	137,9	99.9%	99.5%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POGZ	122,2	99.5%	99.0%	White-Sutton syndrome, 616364
POLG	113,9	100.0%	99.6%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR3A	116,8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090

POLR3B	129,8	99.7%	98.2%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	115,5	100.0%	99.6%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123
POMGNT2	201,7	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	138,7	100.0%	100.0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	130,6	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	103,3	100.0%	98.4%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PORCN	111,2	99.9%	98.8%	Focal dermal hypoplasia, 305600
POU1F1	109,1	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	55,7	84.1%	70.5%	No OMIM phenotype ?Intellectual disability (Dheedene (2014) Mol Syndromol 5,32)
PPM1D	170,2	100.0%	99.6%	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450
PPP1CB	113,1	100.0%	99.1%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	124	100.0%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817

PPP2CA	161	100.0%	99.9%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	129,1	91.6%	91.6%	Mental retardation, autosomal dominant 36, 616362
PPP2R5B	117,2	100.0%	100.0%	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24, 4775)
PPP2R5C	94,3	96.7%	89.1%	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24,4775)
PPP2R5D	136,6	100.0%	100.0%	Mental retardation, autosomal dominant 35, 616355
PPP3CA	121,2	99.6%	97.5%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Epileptic encephalopathy, infantile or early childhood, 1, 617711
PPT1	136,6	90.2%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	163,5	100.0%	100.0%	Renpenning syndrome, 309500
PRKAR1A	79,4	98.6%	92.6%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRMT7	119,1	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	81,8	89.0%	81.7%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	111,6	100.0%	99.9%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRR12	130,7	99.9%	97.5%	No OMIM phenotype
PRRT2	111,8	100.0%	99.0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRSS12	137,8	100.0%	99.6%	Mental retardation, autosomal recessive 1, 249500
PRUNE1	118	100.0%	99.3%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481

PSAP	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	42,8	90.3%	72.5%	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSMD12	85,5	98.5%	90.1%	Stankiewicz-Isidor syndrome, 617516
PSPH	126,6	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PTCH1	110,2	99.9%	98.4%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCHD1	138,5	100.0%	100.0%	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	112	100.0%	99.9%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	129,7	99.6%	97.0%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174
PTF1A	120,6	99.9%	98.2%	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTRH2	200,8	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	189,1	100.0%	99.9%	No OMIM phenotype ?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry)
PTS	101,5	99.8%	98.4%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	163,3	100.0%	99.4%	Verheij syndrome, 615583
PUM1	126,9	100.0%	99.5%	Spinocerebellar ataxia 47, 617931
PURA	207,1	99.5%	96.9%	Mental retardation, autosomal dominant 31, 616158
PUS1	113,3	99.8%	97.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	158,3	100.0%	100.0%	Mental retardation, autosomal recessive 55, 617051

PUS7	135,4	100.0%	99.6%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PYCR1	96	99.7%	97.4%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	116,5	99.7%	96.9%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	129,2	100.0%	99.8%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	97,9	100.0%	99.2%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	133	100.0%	99.4%	Ververi-Brady syndrome, 617982
RAB11B	212,7	100.0%	100.0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	83,4	99.7%	97.2%	Warburg micro syndrome 3, 614222
RAB23	107,4	100.0%	99.2%	Carpenter syndrome, 201000
RAB27A	126,1	100.0%	99.8%	Griscelli syndrome, type 2, 607624
RAB39B	102	100.0%	99.9%	?Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	121,7	99.4%	98.9%	Warburg micro syndrome 1, 600118
RAB3GAP2	91,6	99.7%	96.9%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAC1	101,1	99.6%	94.4%	Mental retardation, autosomal dominant 48, 617751
RAD21	83	97.8%	93.4%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAI1	194,4	100.0%	100.0%	Smith-Magenis syndrome, 182290
RALA	123,4	90.8%	83.6%	No OMIM phenotype
RARB	93,2	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RARS2	104	100.0%	99.4%	Pontocerebellar hypoplasia, type 6, 611523
RBBP8	120,6	99.9%	99.3%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744

RBFOX1	138	89.8%	89.2%	No OMIM phenotype Epilepsy, rolandic (Lal (2013) PLoS One 8, e73323) Mental retardation (Bhalla (2004) J Hum Genet 49, 308 ?Autism spectrum disorder (Griswold (2015) Mol Autism 6, 43) ?Developmental coordination disorder (Mosca (2016) J Med Ge
RBM10	111,6	99.8%	97.6%	TARP syndrome, 311900
RBM28	130,1	100.0%	99.9%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	70,7	96.3%	87.0%	Adams-Oliver syndrome 3, 614814
RCBTB1	95,9	99.8%	99.3%	Retinal dystrophy with or without extraocular anomalies, 617175
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RELN	130,1	100.0%	99.6%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
RERE	77,6	96.5%	93.2%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	137,3	97.6%	97.1%	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6) {Psoriasis,association with} (Strange (2010) Nat Genet 42,985) {Colorectal cancer,increased risk,association with} (Webb (2006) Hum Mol Genet 15,3263)
RFT1	105,7	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015
RHEB	36,8	90.3%	73.3%	No OMIM phenotype
RHOBTB2	190,5	100.0%	99.9%	Epileptic encephalopathy, early infantile, 64, 618004
RIT1	139,2	100.0%	100.0%	Noonan syndrome 8, 615355
RLIM	98	99.5%	97.7%	Tonne-Kalscheuer syndrome, 300978
RMND1	132,6	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	102,2	95.4%	90.2%	Leukoencephalopathy, cystic, without megalencephaly, 612951

RNF113A	134,9	100.0%	100.0%	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	175,4	99.9%	99.2%	Tenorio syndrome, 616260
ROGDI	127,6	100.0%	99.4%	Kohlschutter-Tonz syndrome, 226750
ROR2	160,6	100.0%	99.7%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	103,6	96.8%	91.2%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	65,6	97.1%	86.9%	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPS19	76,7	99.9%	96.6%	Diamond-Blackfan anemia 1, 105650
RPS6KA3	87,8	98.3%	93.0%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	143,9	99.9%	99.4%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RSPRY1	142	100.0%	99.9%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	75,4	99.6%	95.4%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	131,1	99.7%	97.7%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN4IP1	79,6	100.0%	98.0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	117,9	98.8%	97.4%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,3	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	188,1	100.0%	100.0%	Mental retardation, autosomal recessive 61, 617773

SALL1	113,3	99.9%	98.9%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SAMD9	163,9	100.0%	99.9%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS	107,6	99.9%	98.8%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SATB2	107,4	99.8%	97.7%	Glass syndrome, 612313
SBDS	166,2	100.0%	100.0%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SC5D	153,6	99.8%	99.3%	Lathosterolosis, 607330
SCAPER	138,7	98.2%	96.4%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCN1A	121,4	100.0%	99.1%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	169,7	99.9%	98.1%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2A	132,4	99.6%	97.7%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	138,7	99.9%	99.1%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	154,3	100.0%	99.7%	?Myoclonus, familial, 2, 618364 Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SCO1	100,1	99.8%	98.1%	Mitochondrial complex IV deficiency, 220110
SCO2	115,7	100.0%	99.9%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908

SCYL1	146,2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	124,1	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	88,9	85.1%	77.7%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SEMA3E	130,9	100.0%	99.6%	?CHARGE syndrome, 214800
SEPSECS	159,6	100.0%	99.6%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	122,7	98.8%	97.7%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	153,9	99.7%	98.7%	No OMIM phenotype Schizophrenia (Takata (2014) Neuron 82, 723)
SETD1B	171	97.4%	96.4%	No OMIM phenotype
SETD2	137,5	100.0%	99.7%	Luscan-Lumish syndrome, 616831
SETD5	147,6	100.0%	99.7%	Mental retardation, autosomal dominant 23, 615761
SGPL1	132,3	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SGSH	140,2	97.6%	94.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK2	137,1	100.0%	99.8%	{Autism susceptibility 17}, 613436
SHANK3	123,9	97.5%	91.6%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	147,1	100.0%	100.0%	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	139,6	99.9%	99.4%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	96,2	99.9%	98.7%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	118,5	99.6%	96.7%	Epileptic encephalopathy, early infantile, 30, 616341
SIL1	129,5	98.9%	96.2%	Marinesco-Sjogren syndrome, 248800
SIN3A	109,7	99.9%	98.3%	Witteveen-Kolk syndrome, 613406
SIX3	206	100.0%	99.9%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	132,9	100.0%	99.3%	Shprintzen-Goldberg syndrome, 182212

SLC12A5	111,9	86.1%	84.1%	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	118,9	100.0%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	141,9	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	63,3	98.7%	91.0%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	137,7	99.8%	96.1%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	134,6	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	145,6	100.0%	99.6%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A2	97,1	99.3%	97.2%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	146,4	100.0%	99.5%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	103,2	99.3%	95.1%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	150,9	99.9%	99.9%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	146,8	97.9%	93.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	123,8	100.0%	99.1%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	128,9	99.6%	99.1%	Fontaine progeroid syndrome, 612289
SLC2A1	148,9	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC33A1	132	99.7%	97.7%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	127,1	100.0%	99.4%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	104,8	99.8%	98.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	66,6	80.6%	78.3%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	187,8	100.0%	99.8%	Congenital disorder of glycosylation, type IIc, 266265

SLC39A14	95,4	99.9%	97.9%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A8	140,9	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC46A1	111,1	99.9%	98.4%	Folate malabsorption, hereditary, 229050
SLC4A4	113,9	99.8%	98.3%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A1	126	100.0%	100.0%	Myoclonic-atonic epilepsy, 616421
SLC6A17	149,8	100.0%	100.0%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	129,3	100.0%	100.0%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	133	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	53,5	96.1%	83.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	148,8	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	105,5	100.0%	99.6%	Lysinuric protein intolerance, 222700
SLC9A6	101	98.6%	94.3%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	83,6	97.9%	91.3%	Intellectual developmental disorder, X-linked 108, 301024
SMAD4	108,9	100.0%	99.9%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	180,5	98.8%	89.1%	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMARCA1	93,2	99.2%	95.7%	No OMIM phenotype
SMARCA2	105,9	96.8%	95.9%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	150,9	100.0%	99.4%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	179,1	100.0%	99.9%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCC2	98	99.8%	97.9%	Coffin-Siris syndrome 8, 618362

SMARCE1	66,5	94.4%	84.2%	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	87,8	99.9%	97.8%	Cornelia de Lange syndrome 2, 300590
SMC3	84	96.0%	89.7%	Cornelia de Lange syndrome 3, 610759
SMG9	94,5	100.0%	100.0%	Heart and brain malformation syndrome, 616920
SMOC1	115,1	99.8%	98.2%	Microphthalmia with limb anomalies, 206920
SMPD1	146,4	100.0%	99.2%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	63	88.9%	73.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	119	99.9%	99.7%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	168,4	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	131	100.0%	99.2%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRPB	77,7	99.9%	97.6%	Cerebrocostomandibular syndrome, 117650
SNRPN	91,5	100.0%	98.3%	Prader-Willi syndrome, 176270
SNX14	84,1	99.0%	95.4%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	182,7	98.8%	97.8%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	126,8	98.9%	95.2%	ZTTK syndrome, 617140
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	99,7	99.7%	97.9%	Noonan syndrome 9, 616559
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	196,2	100.0%	100.0%	Mental retardation, autosomal dominant 27, 615866
SOX2	230	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	74	97.7%	92.9%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX4	104,5	100.0%	99.2%	No OMIM phenotype
SOX5	89,4	99.7%	97.6%	Lamb-Shaffer syndrome, 616803
SPART	132,6	99.8%	98.2%	Troyer syndrome, 275900

SPAST	95,4	99.8%	97.7%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	139,5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPECC1L	127,5	100.0%	99.8%	?Facial clefting, oblique, 1, 600251 Hypertelorism, Teebi type, 145420 Opitz GBBB syndrome, type II, 145410
SPG11	116,1	99.7%	98.4%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPOCK1	111,1	99.9%	98.8%	No OMIM phenotype Developmental delay and microcephaly (Dhamija (2014) Eur J Med Genet 57,181)
SPR	145,7	100.0%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	146,5	99.8%	98.8%	Legius syndrome, 611431
SPTAN1	112	99.1%	98.3%	Epileptic encephalopathy, early infantile, 5, 613477
SPTBN2	126,2	100.0%	99.7%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SRCAP	153	100.0%	99.6%	Floating-Harbor syndrome, 136140
SRD5A3	139,9	99.8%	98.3%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	61,6	99.4%	93.6%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SSR4	109,5	100.0%	99.7%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	134,7	100.0%	99.5%	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	101,8	89.0%	84.9%	Salt and pepper developmental regression syndrome, 609056
STAG1	111,1	99.6%	97.2%	Mental retardation, autosomal dominant 47, 617635
STAMBP	93,7	99.8%	97.9%	Microcephaly-capillary malformation syndrome, 614261
STIL	154,1	100.0%	99.8%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	117,6	100.0%	99.8%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	108,6	100.0%	98.8%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STT3A	123,1	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127,4	99.9%	99.6%	?Congenital disorder of glycosylation, type Ix, 615597

STX1B	157,7	100.0%	100.0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	103,7	96.8%	96.4%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	58,8	91.7%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	102,9	99.9%	99.6%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
SUOX	167,2	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SURF1	84,8	91.3%	88.4%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SUZ12	106,3	94.6%	87.9%	No OMIM phenotype
SVBP	112,9	100.0%	100.0%	No OMIM phenotype
SYN1	66,6	90.6%	79.1%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	65,9	96.0%	83.8%	No OMIM phenotype ?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)
SYNGAP1	140,7	98.4%	97.7%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	126,6	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	79,9	99.9%	98.1%	Mental retardation, X-linked 96, 300802
SYT1	149,6	99.9%	99.1%	Baker-Gordon syndrome, 618218
SZT2	135,6	99.6%	99.4%	Epileptic encephalopathy, early infantile, 18, 615476
TAF1	86,8	99.1%	95.5%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	100,1	100.0%	99.9%	Mental retardation, autosomal recessive 60, 617432
TAF2	113,9	99.8%	98.5%	Mental retardation, autosomal recessive 40, 615599
TAF6	127,3	100.0%	99.3%	Alazami-Yuan syndrome, 617126
TANC2	134,2	99.9%	99.3%	No OMIM phenotype
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	115	100.0%	100.0%	Tyrosinemia, type II, 276600
TBC1D20	115,7	96.3%	93.8%	Warburg micro syndrome 4, 615663

TBC1D23	92,7	99.2%	95.4%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	177,7	100.0%	100.0%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	99,6	99.8%	99.3%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	136,2	98.2%	94.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	116,4	98.7%	94.7%	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	101,6	99.5%	96.1%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	62,9	94.5%	80.8%	Mental retardation, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBP	99,5	99.9%	99.7%	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBR1	166,2	100.0%	100.0%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TCF20	125,5	100.0%	100.0%	No OMIM phenotype Autism spectrum disorder (Babbs (2014) J Med Genet 51,737)
TCF4	109,2	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	155,1	99.7%	98.0%	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCN2	148,5	100.0%	100.0%	Transcobalamin II deficiency, 275350
TCTN2	122,4	99.9%	99.0%	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860

TDP2	173	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	137,2	100.0%	100.0%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	124,9	100.0%	99.6%	Mental retardation, autosomal recessive 14, 614020
TELO2	122,6	99.8%	97.5%	You-Hoover-Fong syndrome, 616954
TFAP2A	112,7	99.8%	98.0%	Branchiooculofacial syndrome, 113620
TGDS	88,3	99.4%	96.6%	Catel-Manzke syndrome, 616145
TGFBR1	156,4	95.4%	93.8%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGIF1	141,2	100.0%	100.0%	Holoprosencephaly 4, 142946
TH	96,3	100.0%	98.2%	Segawa syndrome, recessive, 605407
THOC2	83,9	98.9%	93.6%	Mental retardation, X-linked 12/35, 300957
THOC6	228,9	100.0%	100.0%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	141,2	99.9%	99.3%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM50	122,9	99.9%	98.7%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	46,3	94.6%	79.9%	Mohr-Tranebjaerg syndrome, 304700
TINF2	177,1	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TKT	115,6	98.7%	98.1%	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	89,3	97.9%	92.3%	Mental retardation, autosomal dominant 57, 618050
TMCO1	81,7	88.0%	87.5%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM165	148,2	100.0%	99.8%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	117,7	100.0%	99.2%	Joubert syndrome 14, 614424
TMEM240	163,9	100.0%	100.0%	Spinocerebellar ataxia 21, 607454
TMEM5	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041

TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	117,9	99.8%	97.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	87,4	99.1%	94.8%	{Autism, susceptibility to, X-linked 6}, 300872
TMTC3	92,8	99.5%	97.6%	Lissencephaly 8, 617255
TNIK	106,3	99.9%	98.7%	Mental retardation, autosomal recessive 54, 617028
TOE1	141,1	100.0%	99.8%	Pontocerebellar hypoplasia, type 7, 614969
TP53RK	81,3	99.7%	96.1%	Galloway-Mowat syndrome 4, 617730
TPI1	112,1	99.9%	96.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	137,6	100.0%	99.6%	Thyroid dyshormonogenesis 2A, 274500
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRKB	59,4	80.3%	73.9%	Galloway-Mowat syndrome 5, 617731
TRAF7	161,7	99.8%	98.1%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	123,4	100.0%	100.0%	Seckel syndrome 9, 616777
TRAPPC11	125,6	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC6B	79	99.9%	98.2%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	125,2	100.0%	99.7%	Mental retardation, autosomal recessive 13, 613192
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIO	121,9	99.0%	96.9%	Mental retardation, autosomal dominant 44, 617061
TRIP12	132,2	99.9%	99.4%	Mental retardation, autosomal dominant 49, 617752

TRIT1	107,9	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	117,1	99.7%	97.6%	Mental retardation, autosomal recessive 68, 618302
TRMT10A	119,5	99.9%	99.2%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRRAP	138	99.7%	99.0%	No OMIM phenotype
TSC1	112,5	99.6%	98.2%	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	140,5	100.0%	99.9%	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	89,8	99.7%	96.4%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	95,6	99.9%	98.9%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	114,4	99.4%	96.8%	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	120	100.0%	99.2%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	229,7	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	107,3	100.0%	99.0%	Mental retardation, X-linked 58, 300210
TTC19	83,4	97.0%	82.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	135,1	99.9%	99.2%	Trichohepatoenteric syndrome 1, 222470
TTC8	115,2	99.8%	98.8%	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	96,2	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	77,6	99.8%	97.1%	Lissencephaly 3, 611603
TUBA8	126,1	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	112,9	98.2%	94.2%	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610

TUBB2A	77,1	99.7%	97.2%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	78,2	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	121,3	99.8%	98.4%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	101,2	97.1%	95.6%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	154,3	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	104,6	98.0%	94.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	155,1	100.0%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	155,1	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TWIST1	160,2	100.0%	99.6%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
TWNK	159,6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
UBA5	79,9	97.7%	86.6%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	117,9	99.9%	96.4%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	81,5	98.6%	93.3%	Angelman syndrome, 105830
UBE3B	113,5	100.0%	99.7%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	119,9	99.8%	99.0%	Johanson-Blizzard syndrome, 243800
UBTF	116,6	100.0%	99.6%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	121,3	100.0%	100.0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	104,9	73.2%	69.6%	Leukodystrophy, hypomyelinating, 14, 617899

UNC13A	122	99.3%	97.3%	No OMIM phenotype
UNC80	111,2	100.0%	99.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	143	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161
UPF3B	61,6	95.3%	84.8%	Mental retardation, X-linked, syndromic 14, 300676
UROC1	132,8	100.0%	99.9%	?Urocanase deficiency, 276880
USP27X	155,1	100.0%	100.0%	Mental retardation, X-linked 105, 300984
USP7	83,4	94.9%	90.1%	No OMIM phenotype ?Autism spectrum disorder (Levy (2011) Neuron 70,886)
USP9X	93,9	98.3%	92.3%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
VAMP1	135,4	100.0%	100.0%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VAMP2	95,7	99.8%	98.9%	No OMIM phenotype
VLDLR	141,4	100.0%	99.9%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	119,6	95.4%	93.1%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
VPS37A	64,3	91.3%	79.3%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	111,3	91.1%	89.6%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	129,6	99.8%	98.7%	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	124,9	99.9%	98.9%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	146,3	100.0%	99.2%	Desanto-Shinawi syndrome, 616708
WARS2	132,3	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	84,2	99.8%	97.8%	No OMIM phenotype
WASHC4	108,9	99.3%	95.9%	?Mental retardation, autosomal recessive 43, 615817
WDPCP	106,7	97.8%	94.9%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR13	116,5	100.0%	99.6%	No OMIM phenotype Intellectual disability,X-linked (Whibley (2010) Am J Hum Genet 87,173)
WDR26	98,2	99.5%	97.4%	Skraban-Deardorff syndrome, 617616

WDR4	142,2	100.0%	100.0%	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45	68,7	96.8%	88.9%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	72,7	97.5%	90.1%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	152,6	100.0%	99.8%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	153,2	100.0%	99.9%	Galloway-Mowat syndrome 1, 251300
WDR81	184,8	100.0%	100.0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WFS1	189,9	100.0%	99.9%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WWOX	116,1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XPA	74,7	99.7%	98.2%	Xeroderma pigmentosum, group A, 278700
XRCC4	143	99.9%	99.0%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	128,1	99.9%	98.2%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YME1L1	103,9	98.2%	93.5%	?Optic atrophy 11, 617302
YWHAE	114,7	100.0%	100.0%	No OMIM phenotype Develomental delay,facial dysmorphology and growth retardation (Enomoto (2012) Am J Med Genet A 158A) Developmental delay and mild brain structural abnormalities (Bi (2009) Nat Genet 41,168)
YWHAG	167,1	100.0%	100.0%	Epileptic encephalopathy, early infantile, 56, 617665
YY1	128,4	100.0%	98.4%	Gabriele-de Vries syndrome, 617557
ZBTB11	164,8	100.0%	99.7%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	148,4	100.0%	100.0%	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447

ZBTB18	177,3	99.9%	99.2%	Mental retardation, autosomal dominant 22, 612337
ZBTB20	180,2	100.0%	100.0%	Primrose syndrome, 259050
ZBTB24	155,5	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	152,9	99.8%	98.5%	Mental retardation, autosomal recessive 56, 617125
ZC4H2	72,4	99.8%	95.9%	Wieacker-Wolff syndrome, 314580
ZDHHC9	48,8	97.7%	87.0%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	140,1	99.7%	98.4%	Mowat-Wilson syndrome, 235730
ZFYVE26	104,8	99.9%	98.7%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	279,6	100.0%	100.0%	Craniosynostosis 6, 616602
ZIC2	165,6	97.5%	95.4%	Holoprosencephaly 5, 609637
ZMIZ1	143,1	99.8%	99.1%	No OMIM phenotype
ZMYND11	119,9	99.9%	99.5%	Mental retardation, autosomal dominant 30, 616083
ZNF148	157,2	100.0%	99.8%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	132,7	99.6%	98.4%	No OMIM phenotype ?Autism (Neale (2012) Nature 485,242)
ZNF407	152,6	100.0%	99.3%	No OMIM phenotype Intellectual disability and autism (Ren (2013) Biochim Biophys Acta 1832,431) Cognitive impairment, failure to thrive, hypotonia and dysmorfic features (Kambouris (2014) Orphanet J Rare Dis 9)
ZNF41	89,6	99.9%	99.3%	Mental retardation, X-linked 89, 300848
ZNF462	156,6	100.0%	99.8%	No OMIM phenotype
ZNF711	114,4	99.7%	98.1%	Mental retardation, X-linked 97, 300803
ZSWIM6	120,9	96.4%	93.7%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.

This list is accurate for panel version DG 2.16

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