INTELLECTUAL DISABILITY GENE PANEL DG 2.17 (1300 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	109.1	100.0%	99.8%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS	109.2	100.0%	99.7%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AASS	131.3	100.0%	99.6%	Hyperlysinemia, 238700
ABAT	86.1	99.9%	98.4%	GABA-transaminase deficiency, 613163
ABCC8	134.7	100.0%	99.9%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	140.4	100.0%	99.9%	Hypertrichotic osteochondrodysplasia, 239850 Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABCD1	95.7	77.7%	74.9%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD4	139.9	99.9%	98.5%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	183.6	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACAD9	130.9	100.0%	98.8%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	164.6	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACAT1	110.0	99.9%	97.1%	Alpha-methylacetoacetic aciduria, 203750
ACO2	125.5	95.6%	90.3%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	129.5	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	158.9	99.9%	99.4%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100.3	98.2%	92.7%	Mental retardation, X-linked 63, 300387
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	131.2	100.0%	100.0%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTL6A	124.1	99.9%	98.6%	No OMIM Disease ID

ACTL6B	144.9	100.0%	100.0%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACVR1	139.8	100.0%	100.0%	Fibrodysplasia ossificans progressiva, 135100
ACY1	128.5	99.9%	99.1%	Aminoacylase 1 deficiency, 609924
ADAM22	133.4	100.0%	99.4%	?Epileptic encephalopathy, early infantile, 61, 617933
ADAR	117.2	99.9%	99.4%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	153.0	100.0%	100.0%	Mental retardation, autosomal recessive 36, 615286
ADGRG1	159.1	100.0%	100.0%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADK	101.3	99.9%	97.3%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	198.1	100.0%	100.0%	Helsmoortel-van der Aa syndrome, 615873
ADPRHL2	177.4	100.0%	100.0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	147.2	99.2%	98.9%	Adenylosuccinase deficiency, 103050
AFF2	109.6	99.9%	98.8%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	102.7	99.9%	98.7%	CHOPS syndrome, 616368
AFG3L2	100.8	95.7%	85.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGO2	128.6	99.2%	99.1%	No OMIM Disease ID
AHCY	120.8	100.0%	98.5%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	172.9	99.9%	98.8%	Xia-Gibbs syndrome, 615829
AHI1	125.5	99.9%	97.6%	Joubert syndrome 3, 608629
AHSG	173.3	100.0%	99.9%	?Alopecia-mental retardation syndrome 1, 203650
AIFM1	92.9	99.7%	96.5%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
AIMP1	80.4	99.1%	91.4%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	126.1	97.0%	89.6%	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	79.2	98.6%	94.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	116.9	100.0%	99.8%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH3A2	116.9	95.3%	94.2%	Sjogren-Larsson syndrome, 270200
ALDH4A1	136.8	100.0%	99.8%	Hyperprolinemia, type II, 239510

ALDH5A1	95.5	99.6%	95.6%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	68.0	94.2%	86.7%	Epilepsy, pyridoxine-dependent, 266100
ALG1	51.3	53.6%	52.1%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	132.1	96.8%	96.5%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	169.5	100.0%	100.0%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	77.7	98.6%	92.4%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG2	112.6	100.0%	100.0%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	117.9	100.0%	100.0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98.1	98.9%	94.9%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118.5	96.8%	95.7%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	114.8	100.0%	99.8%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
ALX3	148.9	91.7%	80.3%	Frontonasal dysplasia 1, 136760
ALX4	175.4	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMER1	106.2	99.8%	99.1%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	102.3	100.0%	99.6%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	146.3	100.0%	100.0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	151.3	100.0%	100.0%	Glycine encephalopathy, 605899
ANK3	144.2	99.4%	99.2%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	116.6	100.0%	100.0%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	155.0	100.0%	99.9%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	131.8	99.6%	97.6%	KBG syndrome, 148050
ANKS1B	122.4	100.0%	99.5%	No OMIM Disease ID
ANO10	106.0	98.6%	96.5%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	112.9	99.3%	97.3%	GAPO syndrome, 230740
AP1S1	105.8	100.0%	99.9%	MEDNIK syndrome, 609313
AP1S2	50.8	75.6%	68.1%	Mental retardation, X-linked syndromic 5, 304340
AP2M1	115.2	100.0%	99.9%	Intellectual developmental disorder 60 with seizures, 618587
AP3B1	108.2	99.4%	95.7%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	135.0	99.8%	97.9%	Epileptic encephalopathy, early infantile, 48, 617276

AP3D1	135.1	98.5%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	124.8	99.9%	98.7%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	103.1	100.0%	99.0%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	140.7	99.9%	98.6%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	66.4	77.7%	70.3%	Spastic paraplegia 52, autosomal recessive, 614067
APC2	142.2	100.0%	99.6%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APOPT1	80.4	82.1%	82.1%	Mitochondrial complex IV deficiency, 220110
APTX	99.2	94.5%	91.6%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	142.5	96.8%	96.6%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARFGEF2	129.2	99.9%	98.9%	Periventricular heterotopia with microcephaly, 608097
ARG1	158.2	100.0%	100.0%	Argininemia, 207800
ARHGAP31	151.5	99.9%	99.1%	Adams-Oliver syndrome 1, 100300
ARHGEF6	112.9	99.0%	94.9%	No OMIM Disease ID
ARHGEF9	52.4	76.3%	72.8%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	145.9	99.7%	98.9%	Coffin-Siris syndrome 2, 614607
ARID1B	150.6	99.5%	99.3%	Coffin-Siris syndrome 1, 135900
ARID2	158.2	99.9%	98.5%	Coffin-Siris syndrome 6, 617808
ARL13B	98.7	100.0%	99.7%	Joubert syndrome 8, 612291
ARL6	91.8	99.9%	97.7%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC9	129.0	100.0%	99.4%	Joubert syndrome 30, 617622
ARSA	154.9	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSE	83.5	98.5%	91.1%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	108.7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	58.2	90.9%	83.3%	Proud syndrome, 300004 Lissencephaly, X-linked 2, 300215 Partington syndrome, 309510 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	124.7	99.6%	96.8%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASH1L	146.8	98.7%	98.4%	Mental retardation, autosomal dominant 52, 617796
ASL	135.7	99.9%	99.2%	Argininosuccinic aciduria, 207900
ASNS	81.9	97.9%	91.0%	Asparagine synthetase deficiency, 615574

ASPA	118.0	99.9%	96.9%	Canavan disease, 271900
ASPM	111.3	99.7%	97.8%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	106.1	95.4%	88.7%	Citrullinemia, 215700
ASXL1	141.0	100.0%	99.6%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	150.0	99.4%	98.2%	Shashi-Pena syndrome, 617190
ASXL3	141.2	99.8%	99.1%	Bainbridge-Ropers syndrome, 615485
ATAD1	63.3	98.8%	91.0%	Hyperekplexia 4, 618011
ATAD3A	100.8	93.8%	88.8%	Harel-Yoon syndrome, 617183
ATIC	114.9	100.0%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	136.1	100.0%	99.4%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATN1	175.9	99.9%	99.2%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP1A1	114.1	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	173.7	100.0%	99.8%	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	173.9	100.0%	100.0%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2A2	150.4	100.0%	99.9%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP2	45.5	89.4%	67.3%	Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP6V0A2	120.5	100.0%	99.6%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	133.1	99.8%	97.3%	Epileptic encephalopathy, infantile or early childhood, 3, 618012 Cutis laxa, autosomal recessive, type IID, 617403
ATP6V1B2	125.1	99.9%	99.1%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	118.4	100.0%	99.7%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268

ATR	142.1	99.9%	98.9%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	86.2	99.1%	95.1%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	135.7	100.0%	99.8%	3-methylglutaconic aciduria, type I, 250950
AUTS2	143.1	99.8%	98.7%	Mental retardation, autosomal dominant 26, 615834
AVPR2	150.2	100.0%	99.8%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
B3GALNT2	94.8	93.1%	91.1%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	96.5	87.5%	80.2%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GLCT	97.3	100.0%	99.7%	Peters-plus syndrome, 261540
B4GALNT1	164.9	99.9%	98.3%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT7	138.9	100.0%	99.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	153.5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAZ2B	129.7	99.9%	98.9%	No OMIM Disease ID
BBS1	156.1	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	156.7	100.0%	100.0%	Bardet-Biedl syndrome 10, 615987
BBS12	193.6	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	153.3	100.0%	99.7%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	113.2	100.0%	98.4%	Bardet-Biedl syndrome 4, 615982
BBS5	94.9	98.4%	92.3%	Bardet-Biedl syndrome 5, 615983
BBS7	136.8	99.0%	95.3%	Bardet-Biedl syndrome 7, 615984
BBS9	113.2	98.8%	94.8%	Bardet-Biedl syndrome 9, 615986
BCAP31	78.0	93.9%	81.2%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	193.6	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	122.4	97.8%	90.2%	Maple syrup urine disease, type Ib, 248600
BCKDK	223.0	100.0%	100.0%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL11A	160.4	99.6%	98.0%	Dias-Logan syndrome, 617101
BCL11B	147.9	100.0%	99.3%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCOR	109.0	99.2%	96.2%	Microphthalmia, syndromic 2, 300166
BCORL1	162.3	99.9%	98.5%	Shukla-Vernon syndrome, 301029
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358

				Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	111.3	99.9%	98.1%	Bloom syndrome, 210900
BOLA3	50.9	99.8%	94.6%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	144.9	96.6%	94.9%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRAT1	155.4	100.0%	99.6%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BRF1	120.2	99.9%	98.8%	Cerebellofaciodental syndrome, 616202
BRPF1	174.8	100.0%	100.0%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	137.2	100.0%	99.1%	No OMIM Disease ID
BRWD3	103.5	98.9%	95.1%	Mental retardation, X-linked 93, 300659
BSCL2	112.9	100.0%	99.9%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	135.6	100.0%	99.8%	Biotinidase deficiency, 253260
BUB1B	121.6	99.9%	99.0%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
C12orf4	126.3	100.0%	99.7%	Mental retardation, autosomal recessive 66, 618221
C12orf57	159.8	100.0%	100.0%	Temtamy syndrome, 218340
C12orf65	112.4	100.0%	99.8%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C2CD3	121.4	95.8%	95.3%	Orofaciodigital syndrome XIV, 615948
C5orf42	122.3	99.7%	97.4%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	141.8	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	99.0	99.9%	97.2%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	112.7	99.3%	96.2%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	101.2	98.2%	96.2%	Spinocerebellar ataxia 6, 183086 Epileptic encephalopathy, early infantile, 42, 617106

				Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	147.4	99.8%	98.6%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	151.7	99.9%	99.4%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	135.3	98.0%	97.8%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	129.3	100.0%	99.5%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1G	165.6	100.0%	99.9%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CAD	147.4	100.0%	99.6%	Epileptic encephalopathy, early infantile, 50, 616457
CAMK2A	125.8	100.0%	99.9%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	122.1	100.0%	100.0%	Mental retardation, autosomal dominant 54, 617799
CAMK2G	114.9	99.9%	99.1%	Mental retardation, autosomal dominant 59, 618522
CAMTA1	197.4	100.0%	99.9%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	158.4	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CARS2	138.8	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	84.5	99.4%	94.0%	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CBL	131.1	97.4%	97.1%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	136.4	100.0%	99.3%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	147.9	100.0%	99.8%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCBE1	80.9	99.8%	98.6%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC115	89.9	88.5%	87.0%	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	126.5	99.3%	96.6%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	105.7	99.0%	95.5%	Ritscher-Schinzel syndrome 2, 300963
CCDC47	144.5	99.0%	96.4%	Trichohepatoneurodevelopmental syndrome, 618268

CCDC88A	90.3	99.3%	96.4%	?PEHO syndrome-like, 617507
CCDC88C	119.2	100.0%	99.7%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCND2	147.6	100.0%	100.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	92.2	91.0%	87.3%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CDC42	89.8	98.1%	90.1%	Takenouchi-Kosaki syndrome, 616737
CDC6	142.2	100.0%	99.7%	?Meier-Gorlin syndrome 5, 613805
CDH11	131.7	100.0%	100.0%	Elsahy-Waters syndrome, 211380
CDH15	173.0	100.0%	99.9%	Mental retardation, autosomal dominant 3, 612580
CDK10	141.9	100.0%	100.0%	Al Kaissi syndrome, 617694
CDK13	133.2	100.0%	99.2%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK5RAP2	109.3	99.8%	99.0%	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	141.7	99.5%	95.5%	No OMIM Disease ID
CDKL5	102.7	95.0%	92.9%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1C	116.5	93.6%	84.7%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDON	110.7	99.9%	99.0%	Holoprosencephaly 11, 614226
CENPF	143.9	99.8%	98.7%	Stromme syndrome, 243605
CENPJ	135.8	100.0%	99.4%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	108.8	99.3%	97.8%	Joubert syndrome 25, 616781
CEP120	131.3	100.0%	99.6%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	88.3	99.1%	92.0%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	145.3	99.6%	97.8%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	77.6	96.9%	88.7%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	79.1	98.7%	94.4%	Joubert syndrome 15, 614464
CEP57	84.9	99.3%	92.8%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP83	103.3	99.8%	96.2%	Nephronophthisis 18, 615862
CEP89	130.0	98.2%	95.5%	No OMIM disease ID
CHAMP1	181.4	100.0%	100.0%	Mental retardation, autosomal dominant 40, 616579
CHD1	104.2	97.7%	90.7%	Pilarowski-Bjornsson syndrome, 617682

CHD2	126.1	99.4%	99.1%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	101.6	98.4%	95.1%	Snijders Blok-Campeau syndrome, 618205
CHD4	117.3	100.0%	99.9%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	143.6	100.0%	99.5%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	133.1	100.0%	99.9%	No OMIM disease ID
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	133.0	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
CHRNA4	125.3	100.0%	99.3%	Epilepsy, nocturnal frontal lobe, 1, 600513
CIC	83.4	64.8%	63.4%	Mental retardation, autosomal dominant 45, 617600
CIT	106.3	100.0%	98.9%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	154.2	99.9%	98.9%	Filippi syndrome, 272440
CLCN4	111.7	99.9%	98.8%	Raynaud-Claes syndrome, 300114
CLIC2	72.0	100.0%	98.2%	?Mental retardation, X-linked, syndromic 32, 300886
CLIP1	122.0	99.9%	99.1%	No OMIM Disease ID
CLN3	123.4	92.5%	92.2%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.4	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	141.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	156.2	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	146.0	100.0%	100.0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	135.3	99.7%	97.4%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLTC	154.5	100.0%	99.8%	Mental retardation, autosomal dominant 56, 617854
CNKSR2	88.5	98.6%	92.0%	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	222.5	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNOT1	127.6	100.0%	99.8%	Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	130.4	100.0%	99.8%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	162.6	100.0%	100.0%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNPY3	83.9	100.0%	100.0%	Epileptic encephalopathy, early infantile, 60, 617929
CNTNAP2	129.8	100.0%	99.8%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COASY	190.6	100.0%	100.0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COG1	117.9	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209

COG4	99.0	100.0%	99.7%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	123.1	99.8%	97.9%	Congenital disorder of glycosylation, type IIi, 613612
COG6	87.3	98.6%	95.6%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	111.8	100.0%	99.9%	Congenital disorder of glycosylation, type IIe, 608779
COG8	160.1	100.0%	98.6%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	100.3	99.8%	98.0%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A2	118.1	100.0%	99.4%	Brain small vessel disease 2, 614483
COL4A3BP	132.6	99.6%	97.3%	Mental retardation, autosomal dominant 34, 616351
COLEC11	197.8	100.0%	100.0%	3MC syndrome 2, 265050
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	116.2	91.7%	90.8%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	177.7	100.0%	100.0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	78.6	99.9%	98.5%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	232.8	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX15	90.4	99.9%	98.7%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	143.0	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
CPLX1	119.2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 63, 617976
CPS1	133.4	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300
CRADD	120.5	100.0%	99.0%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	122.9	88.2%	87.4%	Mental retardation, autosomal recessive 2, 607417
CREBBP	120.9	99.6%	97.3%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRLF1	140.3	93.9%	91.4%	Cold-induced sweating syndrome 1, 272430
CSNK2A1	106.2	94.0%	89.7%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	136.5	100.0%	100.0%	No OMIM Disease ID
CSPP1	117.4	100.0%	99.4%	Joubert syndrome 21, 615636
CSTB	74.8	99.1%	93.0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	111.3	95.7%	88.6%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	131.3	100.0%	99.1%	Mental retardation, autosomal dominant 21, 615502

CTDP1	141.7	96.2%	88.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	110.0	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNB1	129.5	100.0%	100.0%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
CTNND2	104.9	97.9%	93.9%	No OMIM Disease ID
CTSA	146.1	100.0%	100.0%	Galactosialidosis, 256540
CTSD	187.3	100.0%	99.0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	119.0	99.7%	97.9%	No OMIM Disease ID
CUL4B	75.8	97.7%	88.2%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX1	128.7	99.0%	96.5%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	134.5	100.0%	99.4%	Epileptic encephalopathy, early infantile, 67, 618141
CWC27	82.5	99.8%	97.3%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	103.5	100.0%	99.1%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXorf56	74.7	99.5%	93.2%	?Mental retardation, X-linked 107, 301013
CYB5R3	163.2	99.6%	98.5%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYFIP2	122.2	99.9%	98.7%	Epileptic encephalopathy, early infantile, 65, 618008
CYP27A1	184.4	100.0%	99.8%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	139.8	99.1%	96.8%	Spastic paraplegia 56, autosomal recessive, 615030
D2HGDH	157.7	100.0%	99.8%	D-2-hydroxyglutaric aciduria, 600721
DAG1	205.3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DARS	121.4	99.9%	99.0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	125.4	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	109.6	99.7%	96.9%	Maple syrup urine disease, type II, 248600
DCAF17	87.5	100.0%	99.2%	Woodhouse-Sakati syndrome, 241080
DCC	119.4	100.0%	99.9%	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500
DCHS1	164.5	100.0%	100.0%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390

DCPS	140.6	100.0%	99.9%	Al-Raqad syndrome, 616459
DCX	93.7	99.8%	98.9%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDC	100.3	99.4%	96.2%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	130.8	100.0%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	108.3	88.3%	82.2%	Warsaw breakage syndrome, 613398
DDX3X	74.5	86.2%	82.8%	Mental retardation, X-linked 102, 300958
DDX59	143.3	100.0%	99.7%	Orofaciodigital syndrome V, 174300
DDX6	58.0	95.7%	81.6%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	124.1	100.0%	99.1%	Mental retardation, autosomal dominant 24, 615828 ?Dyskinesia, seizures, and intellectual developmental disorder, 617171
DEGS1	150.2	100.0%	100.0%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	101.5	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	129.9	100.0%	99.8%	Epilepsy, familial focal, with variable foci 1, 604364
DHCR24	170.7	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	84.5	97.3%	94.0%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DHFR	48.6	92.6%	80.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	126.4	100.0%	100.0%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	127.4	99.9%	99.0%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	178.6	100.0%	100.0%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DIAPH1	104.4	100.0%	99.8%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIP2B	131.4	100.0%	100.0%	Mental retardation, FRA12A type, 136630
DIS3L2	151.0	100.0%	99.9%	Perlman syndrome, 267000
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DLD	117.2	100.0%	99.9%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	84.8	99.3%	94.6%	Mental retardation, X-linked 90, 300850
DLG4	158.8	100.0%	100.0%	No OMIM Disease ID
DMD	107.5	99.6%	97.9%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DMPK	166.2	100.0%	99.9%	Myotonic dystrophy 1, 160900
DNAJC12	144.7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384

DNAJC19	92.5	99.1%	90.4%	3-methylglutaconic aciduria, type V, 610198
DNM1	151.2	94.9%	93.3%	Epileptic encephalopathy, early infantile, 31, 616346
DNMT3A	132.4	99.9%	98.7%	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	125.5	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK3	118.4	100.0%	99.5%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	132.3	99.6%	98.9%	Adams-Oliver syndrome 2, 614219
DOCK7	118.3	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DOLK	171.4	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DONSON	92.4	99.6%	94.7%	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPAGT1	93.2	100.0%	100.0%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	105.3	99.9%	97.4%	Coffin-Siris syndrome 7, 618027
DPH1	177.3	100.0%	100.0%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	134.2	95.5%	87.7%	Congenital disorder of glycosylation, type Ie, 608799
DPP6	129.8	100.0%	99.8%	Mental retardation, autosomal dominant 33, 616311
DPYD	140.7	99.4%	96.2%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	121.4	100.0%	99.9%	Dihydropyrimidinuria, 222748
DYM	102.4	97.4%	95.7%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	149.0	100.0%	99.8%	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
DYNC1I2	49.4	83.2%	67.5%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYRK1A	134.0	100.0%	100.0%	Mental retardation, autosomal dominant 7, 614104
EBF3	153.4	100.0%	99.9%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	68.9	99.8%	96.3%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	111.6	100.0%	100.0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDC3	120.5	100.0%	99.2%	?Mental retardation, autosomal recessive 50, 616460
EED	83.9	98.8%	93.6%	Cohen-Gibson syndrome, 617561
EEF1A2	209.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFNB2	151.8	100.0%	99.6%	No OMIM Disease ID
EFTUD2	107.1	100.0%	99.5%	Mandibulofacial dysostosis, Guion-Almeida type, 610536

EHMT1	138.4	94.7%	94.5%	Kleefstra syndrome 1, 610253
EIF2AK3	134.0	99.5%	96.7%	Wolcott-Rallison syndrome, 226980
EIF2S3	82.9	96.7%	88.2%	MEHMO syndrome, 300148
EIF3F	67.7	99.3%	92.9%	Mental retardation, autosomal recessive 67, 618295
EIF4A3	89.6	100.0%	98.9%	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	117.1	100.0%	99.5%	Combined oxidative phosphorylation deficiency 17, 615440
ELOVL4	103.3	100.0%	99.6%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP2	118.6	99.8%	97.6%	Mental retardation, autosomal recessive 58, 617270
EMC1	111.0	100.0%	99.2%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EML1	134.8	100.0%	99.9%	Band heterotopia, 600348
EMX2	175.9	100.0%	100.0%	Schizencephaly, 269160
ENTPD1	125.9	100.0%	100.0%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	173.3	99.7%	98.7%	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
EPG5	111.4	99.5%	98.3%	Vici syndrome, 242840
ERCC1	91.5	100.0%	98.8%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	139.5	100.0%	99.9%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	95.9	99.9%	98.7%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC5	130.9	100.0%	99.4%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	161.8	100.0%	100.0%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ERCC8	79.9	99.0%	89.3%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	119.3	100.0%	99.4%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	112.4	99.5%	96.2%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000

ETFB	127.7	100.0%	100.0%	Glutaric acidemia IIB, 231680
ETHE1	105.9	99.9%	97.9%	Ethylmalonic encephalopathy, 602473
EXOSC2	114.1	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	135.7	96.5%	87.0%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC9	133.4	99.2%	95.0%	Pontocerebellar hypoplasia, type 1D, 618065
EXTL3	200.7	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	130.3	99.5%	98.0%	Weaver syndrome, 277590
FA2H	101.5	99.3%	95.1%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	124.1	100.0%	98.9%	Leukodystrophy, hypomyelinating, 5, 610532
FAM20C	165.1	100.0%	100.0%	Raine syndrome, 259775
FAR1	72.4	97.6%	91.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	169.5	100.0%	100.0%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	77.5	96.9%	93.0%	Rajab interstitial lung disease with brain calcifications, 613658
FAT4	195.5	100.0%	100.0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBXL3	187.9	100.0%	100.0%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	165.9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	82.9	98.5%	94.3%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	123.6	100.0%	98.7%	?Mental retardation, autosomal recessive 45, 615979
FGD1	93.2	98.7%	94.4%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	105.7	100.0%	100.0%	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	225.9	100.0%	100.0%	Spinocerebellar ataxia 27, 609307
FGFR1	131.6	100.0%	99.7%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	118.0	97.7%	97.1%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592

				LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	157.1	100.0%	99.9%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FH	126.0	95.9%	89.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	135.9	100.0%	100.0%	Thauvin-Robinet-Faivre syndrome, 617107
FIGN	141.0	100.0%	100.0%	No OMIM Disease ID
FKRP	178.0	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	108.0	99.9%	96.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLNA	156.4	100.0%	99.9%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350

				Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FLVCR1	154.8	100.0%	99.4%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	131.7	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	120.8	87.1%	84.7%	Mental retardation, autosomal recessive 47, 616193
FMR1	78.1	96.3%	90.1%	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624
FOLR1	115.7	100.0%	100.0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	162.5	99.7%	96.6%	Rett syndrome, congenital variant, 613454
FOXP1	117.6	100.0%	99.9%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	130.1	99.5%	98.4%	Speech-language disorder-1, 602081
FOXRED1	129.1	99.9%	99.0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	123.1	99.9%	99.3%	Fraser syndrome 1, 219000
FRMD4A	124.0	91.5%	91.0%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMPD4	115.6	99.7%	97.7%	Mental retardation, X-linked 104, 300983
FRRS1L	100.0	89.3%	81.8%	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	129.8	99.1%	96.1%	Glutamate formiminotransferase deficiency, 229100
FTO	99.1	83.8%	83.7%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	134.4	99.6%	96.5%	Mental retardation, X-linked 9/44, 309549
FUCA1	135.9	100.0%	100.0%	Fucosidosis, 230000
FUT8	130.4	100.0%	99.4%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
GABBR2	115.6	99.1%	95.6%	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 Epileptic encephalopathy, early infantile, 59, 617904
GABRA1	164.8	100.0%	100.0%	Epileptic encephalopathy, early infantile, 19, 615744
GABRA3	84.1	99.2%	95.7%	No OMIM Disease ID
GABRB1	174.8	100.0%	100.0%	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	134.8	100.0%	100.0%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	139.5	99.7%	98.5%	Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	126.9	91.0%	90.0%	Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAD1	114.6	100.0%	99.8%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	103.0	99.8%	98.1%	Krabbe disease, 245200

GALE	153.0	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALT	165.3	100.0%	100.0%	Galactosemia, 230400
GAMT	125.7	99.7%	94.3%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	101.2	100.0%	99.6%	Mental retardation, autosomal dominant 18, 615074
GATM	139.0	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	91.0	99.9%	99.4%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCSH	32.0	94.3%	74.1%	?Glycine encephalopathy, 605899
GDI1	145.2	99.4%	97.9%	Mental retardation, X-linked 41, 300849
GFAP	111.6	91.9%	91.7%	Alexander disease, 203450
GFM1	104.4	100.0%	98.9%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	117.8	99.0%	95.2%	Combined oxidative phosphorylation deficiency 39, 618397
GJA1	162.4	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJB1	161.4	100.0%	100.0%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	59.7	97.7%	86.5%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GK	43.3	82.3%	61.8%	Glycerol kinase deficiency, 307030
GLB1	87.4	99.5%	95.2%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	60.8	91.8%	80.4%	Glycine encephalopathy, 605899
GLI2	177.4	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	151.8	100.0%	99.5%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS3	133.1	100.0%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLUD1	66.3	98.0%	88.9%	Hyperinsulinism-hyperammonemia syndrome, 606762

GLYCTK	175.0	100.0%	99.8%	D-glyceric aciduria, 220120
GM2A	129.1	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMPPA	158.4	100.0%	100.0%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	233.1	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	160.9	93.8%	93.8%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAS	241.4	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GNB1	148.9	100.0%	100.0%	Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GNB5	119.4	99.9%	98.0%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	128.8	99.7%	96.6%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	149.5	100.0%	99.4%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	199.0	99.9%	99.4%	Mucolipidosis III gamma, 252605
GNS	93.6	99.9%	97.2%	Mucopolysaccharidosis type IIID, 252940
GPAA1	137.4	99.9%	98.9%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	76.8	98.9%	93.5%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC4	107.1	99.9%	97.7%	Keipert syndrome, 301026
GPHN	147.8	99.8%	98.8%	Molybdenum cofactor deficiency C, 615501
GPSM2	121.0	100.0%	99.4%	Chudley-McCullough syndrome, 604213
GPT2	130.4	100.0%	99.6%	Mental retardation, autosomal recessive 49, 616281
GRIA3	83.0	98.9%	93.2%	Mental retardation, X-linked 94, 300699
GRIA4	123.6	99.6%	97.9%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	148.3	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	121.6	96.2%	95.3%	Mental retardation, autosomal recessive, 6, 611092

GRIN1	186.6	100.0%	100.0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	139.8	100.0%	100.0%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	168.9	99.9%	99.2%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	92.8	96.7%	85.3%	Epileptic encephalopathy, early infantile, 46, 617162
GRIP1	114.2	100.0%	99.4%	Fraser syndrome 3, 617667
GRM1	167.3	100.0%	100.0%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRN	193.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GSE1	135.4	100.0%	99.9%	No OMIM Disease ID
GSS	98.9	100.0%	99.6%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	82.0	99.8%	97.4%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	138.7	99.9%	99.2%	Jaberi-Elahi syndrome, 617988
GTPBP3	189.1	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	106.6	92.6%	91.1%	Mucopolysaccharidosis VII, 253220
HACE1	135.7	100.0%	99.4%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	118.2	99.3%	99.2%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	74.6	96.1%	89.6%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HAX1	146.3	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	90.4	99.6%	96.6%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	114.5	99.5%	96.5%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	144.1	100.0%	99.9%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	131.1	100.0%	100.0%	No OMIM Disease ID
HDAC6	122.8	99.8%	98.0%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	110.1	100.0%	99.6%	Cornelia de Lange syndrome 5, 300882
HECW2	112.4	99.9%	98.8%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268

HEPACAM	127.8	95.0%	89.6%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	145.0	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	99.7	80.6%	76.7%	Mental retardation, autosomal recessive 38, 615516
HESX1	65.7	99.9%	97.5%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	112.3	93.8%	92.6%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	173.2	99.8%	97.3%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	99.9	88.2%	86.3%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	69.7	95.5%	75.9%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIST1H1E	125.1	100.0%	100.0%	Rahman syndrome, 617537
HIST1H4C	109.9	100.0%	100.0%	No OMIM Disease ID
HIVEP2	171.1	100.0%	100.0%	Mental retardation, autosomal dominant 43, 616977
HLCS	148.0	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMGCL	124.9	100.0%	99.5%	HMG-CoA lyase deficiency, 246450
HNMT	132.7	100.0%	99.7%	Mental retardation, autosomal recessive 51, 616739
HNRNPH2	131.4	100.0%	100.0%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	61.0	87.9%	78.3%	Au-Kline syndrome, 616580
HNRNPU	154.7	100.0%	99.3%	Epileptic encephalopathy, early infantile, 54, 617391
HOXA1	184.7	100.0%	100.0%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HPD	159.7	100.0%	99.9%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	56.9	97.8%	87.8%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HRAS	196.0	100.0%	100.0%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HSD17B10	98.0	100.0%	99.5%	HSD10 mitochondrial disease, 300438

HSD17B4	106.4	95.5%	93.1%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPA9	83.8	88.2%	84.2%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	73.7	97.9%	92.1%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	145.3	100.0%	99.7%	3-methylglutaconic aciduria, type VIII, 617248
HUWE1	82.1	99.1%	95.0%	Mental retardation, X-linked syndromic, Turner type, 309590
HYLS1	160.4	100.0%	100.0%	Hydrolethalus syndrome, 236680
IARS	124.2	99.9%	99.2%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	145.7	100.0%	100.0%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IDS	105.1	99.8%	97.2%	Mucopolysaccharidosis II, 309900
IDUA	169.2	99.3%	96.4%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IER3IP1	108.7	88.3%	80.0%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	110.9	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT81	90.5	93.0%	88.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	106.4	98.8%	93.9%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	100.6	100.0%	100.0%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	123.1	100.0%	99.7%	Insulin-like growth factor I, resistance to, 270450
IKBKG	64.7	90.1%	80.2%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IL1RAPL1	100.0	99.5%	97.6%	Mental retardation, X-linked 21/34, 300143
IMPA1	73.2	96.0%	84.6%	Mental retardation, autosomal recessive 59, 617323
INPP5E	131.1	100.0%	99.3%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPP5K	94.7	100.0%	99.5%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
IQSEC2	81.5	96.6%	90.5%	Mental retardation, X-linked 1/78, 309530

IRF2BPL	197.8	99.6%	97.8%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ISCA2	112.2	99.8%	97.4%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISPD	112.0	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITGA7	142.3	99.8%	98.4%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	142.5	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647
ITPR1	136.4	100.0%	99.8%	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700
IVD	106.7	100.0%	100.0%	Isovaleric acidemia, 243500
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
JAM3	132.0	100.0%	100.0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	136.5	99.9%	99.5%	No OMIM Disease ID
KANK1	129.0	100.0%	99.7%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	156.7	99.8%	99.1%	Koolen-De Vries syndrome, 610443
KAT6A	155.1	100.0%	99.9%	Mental retardation, autosomal dominant 32, 616268
KAT6B	162.3	99.8%	99.2%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KATNB1	170.5	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNA2	132.6	100.0%	99.4%	Epileptic encephalopathy, early infantile, 32, 616366
KCNA4	128.0	100.0%	100.0%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNB1	141.6	100.0%	99.8%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	189.4	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	126.1	94.7%	80.0%	Spinocerebellar ataxia 13, 605259
KCNH1	159.4	98.7%	98.4%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	157.5	89.3%	88.6%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	222.1	100.0%	100.0%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ6	165.7	100.0%	100.0%	Keppen-Lubinsky syndrome, 614098
KCNK4	221.2	100.0%	100.0%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381

KCNK9	189.3	100.0%	100.0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	133.0	91.5%	90.4%	Epileptic encephalopathy, early infantile, 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	116.5	100.0%	98.7%	Seizures, benign neonatal, 2, 121201
KCNQ5	140.1	99.7%	98.5%	Mental retardation, autosomal dominant 46, 617601
KCNT1	145.3	96.3%	95.2%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	171.1	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	134.2	100.0%	99.4%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	124.1	99.7%	97.9%	No OMIM Disease ID
KDM5B	123.2	99.1%	97.2%	Mental retardation, autosomal recessive 65, 618109
KDM5C	111.1	99.6%	97.5%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	98.1	95.6%	87.7%	Kabuki syndrome 2, 300867
KDM6B	160.4	99.9%	98.1%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KIAA0586	115.1	97.3%	92.6%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1109	137.8	99.8%	98.9%	Alkuraya-Kucinskas syndrome, 617822
KIDINS220	138.5	100.0%	99.9%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	89.7	97.6%	94.9%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	112.4	99.8%	97.7%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	125.3	99.8%	98.2%	Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255 Spastic paraplegia 30, autosomal recessive, 610357
KIF1BP	168.4	96.1%	96.1%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF2A	103.8	99.5%	95.9%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	78.5	98.5%	93.1%	?Mental retardation, X-linked 100, 300923
KIF5C	113.8	100.0%	99.0%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL3	138.0	99.9%	99.4%	No OMIM Disease ID
KLF7	131.4	100.0%	100.0%	No OMIM Disease ID
KLHL15	143.1	100.0%	99.9%	Mental retardation, X-linked 103, 300982
KMT2A	138.5	100.0%	99.9%	Wiedemann-Steiner syndrome, 605130

KMT2B	156.7	98.1%	94.7%	Dystonia 28, childhood-onset, 617284
KMT2C	142.7	91.8%	90.3%	Kleefstra syndrome 2, 617768
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
KMT2E	156.1	99.9%	98.4%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	170.8	100.0%	99.9%	Mental retardation, autosomal dominant 51, 617788
KNL1	103.6	99.0%	97.3%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	163.9	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
L1CAM	138.3	99.9%	98.6%	MASA syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000
L2HGDH	123.6	99.2%	97.2%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	119.9	100.0%	99.6%	Poretti-Boltshauser syndrome, 615960
LAMA2	131.6	100.0%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMB1	147.7	100.0%	99.7%	Lissencephaly 5, 615191
LAMC3	163.5	99.9%	99.2%	Cortical malformations, occipital, 614115
LAMP2	89.8	97.8%	92.3%	Danon disease, 300257
LARGE1	122.8	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	74.1	89.4%	74.5%	Alazami syndrome, 615071
LAS1L	83.2	99.5%	96.4%	Wilson-Turner syndrome, 309585
LIAS	124.4	100.0%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	170.5	100.0%	99.9%	LIG4 syndrome, 606593

LINGO1	231.7	100.0%	100.0%	Mental retardation, autosomal recessive 64, 618103
LINS1	135.6	100.0%	99.5%	Mental retardation, autosomal recessive 27, 614340
LMAN2L	113.2	100.0%	99.4%	?Mental retardation, autosomal recessive, 52, 616887
LONP1	164.8	100.0%	100.0%	CODAS syndrome, 600373
LRP2	140.5	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRPPRC	126.3	100.0%	99.7%	Leigh syndrome, French-Canadian type, 220111
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
LZTFL1	116.5	99.9%	99.2%	Bardet-Biedl syndrome 17, 615994
LZTR1	157.2	100.0%	99.9%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAB21L1	187.7	100.0%	100.0%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	265.2	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	133.2	100.0%	99.5%	Lissencephaly 9 with complex brainstem malformation, 618325
MAF	103.8	89.3%	84.7%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAG	175.3	100.0%	99.9%	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	137.7	98.8%	94.4%	Schaaf-Yang syndrome, 615547
MAN1B1	137.5	100.0%	99.9%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	139.1	99.9%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MANBA	117.1	99.7%	98.1%	Mannosidosis, beta, 248510
MAOA	100.1	100.0%	99.2%	Brunner syndrome, 300615
MAP1B	131.5	99.9%	99.6%	No OMIM Disease ID
MAP2K1	96.7	99.6%	97.1%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	139.8	99.3%	95.6%	Cardiofaciocutaneous syndrome 4, 615280
MAPK8IP3	181.0	100.0%	100.0%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPRE2	163.4	99.8%	98.4%	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	140.7	100.0%	99.5%	3MC syndrome 1, 257920
MAST1	187.1	100.0%	99.9%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAT1A	154.2	99.7%	98.2%	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MBD5	151.8	99.9%	99.8%	Mental retardation, autosomal dominant 1, 156200
MBOAT7	121.9	100.0%	99.9%	Mental retardation, autosomal recessive 57, 617188
MBTPS2	109.0	99.9%	98.4%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800

MCCC1	138.0	100.0%	99.6%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	121.8	100.0%	99.9%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	170.5	100.0%	99.4%	Mucolipidosis IV, 252650
MCPH1	138.1	100.0%	98.7%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	116.5	98.0%	98.0%	Epileptic encephalopathy, early infantile, 51, 617339
MECP2	135.2	100.0%	99.5%	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	114.2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	89.4	99.6%	96.5%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MED13	146.3	100.0%	99.7%	No OMIM Disease ID
MED13L	112.2	100.0%	99.8%	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MED17	134.7	97.8%	94.7%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	131.6	99.7%	98.5%	Mental retardation, autosomal recessive 18, 614249
MED25	148.0	100.0%	99.9%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	131.2	99.5%	95.7%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	158.7	100.0%	99.8%	Carpenter syndrome 2, 614976
MEIS2	128.4	100.0%	99.9%	Cleft palate, cardiac defects, and mental retardation, 600987
METTL23	117.0	100.0%	100.0%	Mental retardation, autosomal recessive 44, 615942
METTL5	107.2	99.3%	97.0%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	86.8	93.5%	88.5%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	121.3	100.0%	99.6%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	117.4	100.0%	99.6%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	155.6	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066
MGP	134.2	98.6%	93.2%	Keutel syndrome, 245150
MICU1	103.4	98.7%	96.2%	Myopathy with extrapyramidal signs, 615673
MID1	131.5	99.9%	98.4%	Opitz GBBB syndrome, type I, 300000
MID2	111.8	99.7%	98.0%	?Mental retardation, X-linked 101, 300928

MKKS	161.5	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	98.8	99.9%	98.5%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	102.4	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	105.5	99.7%	97.3%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168.7	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.3	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	214.4	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	76.8	93.0%	77.2%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	101.3	99.3%	96.6%	Molybdenum cofactor deficiency A, 252150
MOCS2	134.2	99.6%	99.6%	Molybdenum cofactor deficiency B, 252160
MOGS	157.9	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	110.1	100.0%	99.8%	Congenital disorder of glycosylation, type If, 609180
MPDZ	127.3	99.6%	98.1%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPLKIP	106.6	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRPS22	138.5	100.0%	97.3%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MSL3	77.3	95.2%	84.1%	Mental retardation, X-linked, syndromic, 36, 301032
MSMO1	47.0	96.2%	87.7%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTFMT	131.5	100.0%	99.7%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	124.2	98.5%	96.7%	Homocystinuria due to MTHFR deficiency, 236250
MTOR	116.6	100.0%	99.2%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTR	134.7	100.0%	99.6%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	135.6	100.0%	99.2%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MUT	128.8	100.0%	99.0%	Methylmalonic aciduria, mut(0) type, 251000
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYCN	200.6	100.0%	100.0%	Feingold syndrome 1, 164280

MYH9	140.9	99.7%	99.0%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	109.9	99.7%	98.5%	Griscelli syndrome, type 1, 214450
MYT1L	154.1	100.0%	99.9%	Mental retardation, autosomal dominant 39, 616521
NAA10	112.8	100.0%	99.4%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAA15	92.0	97.6%	94.4%	Mental retardation, autosomal dominant 50, 617787
NACC1	188.0	100.0%	100.0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAGA	131.4	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	130.5	98.5%	95.6%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NALCN	117.7	99.7%	98.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANS	105.0	100.0%	99.3%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	121.5	97.6%	97.4%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAXE	90.7	99.8%	97.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	121.3	91.9%	90.4%	No OMIM Disease ID
NBN	90.6	100.0%	98.2%	Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065
NDE1	95.8	100.0%	99.8%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDP	96.2	100.0%	99.6%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	204.8	100.0%	100.0%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	195.4	99.9%	98.7%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	129.7	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	162.1	100.0%	99.9%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	179.9	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFAF3	159.3	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	128.0	99.8%	99.3%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFS1	140.7	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	102.9	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	132.3	90.7%	90.6%	Mitochondrial complex I deficiency, nuclear type 8, 618230

NDUFS4	148.6	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	120.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	157.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	171.3	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	154.4	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	71.5	91.8%	77.9%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	96.3	72.4%	71.5%	Periventricular nodular heterotopia 7, 617201
NEU1	150.1	99.5%	96.5%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	135.1	99.9%	99.4%	Mental retardation, X-linked 98, 300912
NF1	105.8	92.5%	89.3%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210
NFIA	158.0	99.9%	99.0%	Brain malformations with or without urinary tract defects, 613735
NFIB	109.8	97.5%	96.8%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	195.4	100.0%	99.8%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFU1	61.6	96.2%	77.8%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	134.1	100.0%	99.9%	Congenital disorder of deglycosylation, 615273
NHS	114.3	98.6%	96.5%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIPBL	123.0	98.9%	96.7%	Cornelia de Lange syndrome 1, 122470
NKX2-1	102.8	100.0%	99.9%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN3	129.8	100.0%	99.2%	No OMIM disease ID
NLGN4X	154.6	99.5%	97.8%	Mental retardation, X-linked, 300495
NONO	83.1	99.7%	96.8%	Mental retardation, X-linked, syndromic 34, 300967
NPC1	120.3	100.0%	99.4%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	130.7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPHP1	119.7	99.8%	97.8%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NR2F1	261.9	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	153.2	100.0%	100.0%	No OMIM Disease ID

NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NRXN1	147.9	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	152.6	100.0%	99.8%	Sotos syndrome 1, 117550
NSD2	135.1	99.9%	98.3%	No OMIM Disease ID
NSDHL	133.7	99.9%	98.2%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	96.2	98.5%	94.8%	Mental retardation, autosomal recessive 5, 611091
NT5C2	119.4	97.9%	95.8%	Spastic paraplegia 45, autosomal recessive, 613162
NTRK1	144.9	100.0%	99.7%	Medullary thyroid carcinoma, familial, 155240 Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	142.9	100.0%	99.9%	Obesity, hyperphagia, and developmental delay, 613886 Epileptic encephalopathy, early infantile, 58, 617830
NUBPL	102.4	98.9%	94.2%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP62	124.5	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
NUS1	51.9	72.8%	44.9%	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	69.1	80.2%	69.8%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	179.8	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OCRL	106.2	99.9%	98.6%	Lowe syndrome, 309000 Dent disease 2, 300555
ODC1	123.0	100.0%	98.7%	No OMIM disease ID
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGT	108.0	99.9%	98.7%	Mental retardation, X-linked 106, 300997
OPHN1	80.7	99.0%	95.5%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	93.8	99.9%	98.9%	Meier-Gorlin syndrome 1, 224690
OSGEP	104.4	100.0%	97.8%	Galloway-Mowat syndrome 3, 617729
OTC	111.3	100.0%	99.7%	Ornithine transcarbamylase deficiency, 311250

OTUD6B	118.1	99.9%	97.9%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTX2	135.4	100.0%	99.6%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P4HTM	177.7	100.0%	99.4%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PACS1	115.1	100.0%	99.8%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	170.7	100.0%	99.6%	Epileptic encephalopathy, early infantile, 66, 618067
PAFAH1B1	78.7	91.7%	82.7%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	128.9	100.0%	100.0%	Phenylketonuria, 261600
PAK1	102.7	100.0%	99.3%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	85.7	99.0%	94.4%	Mental retardation, X-linked 30/47, 300558
PANK2	161.5	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	137.7	100.0%	99.9%	Oocyte maturation defect 7, 618550
PARN	127.9	100.0%	99.6%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX1	212.4	98.6%	94.1%	?Otofaciocervical syndrome 2, 615560
PAX6	122.8	100.0%	99.9%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Anterior segment dysgenesis 5, multiple subtypes, 604229
PAX7	147.9	100.0%	100.0%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX8	103.2	100.0%	100.0%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	115.4	100.0%	98.3%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	170.2	99.9%	98.8%	Pyruvate carboxylase deficiency, 266150
PCCA	97.7	99.1%	95.4%	Propionicacidemia, 606054
PCCB	114.9	99.5%	97.1%	Propionicacidemia, 606054
PCDH12	195.1	100.0%	100.0%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280

PCDH19	192.4	99.9%	98.9%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	103.7	99.8%	97.3%	Turnpenny-Fry syndrome, 618371
PCLO	147.8	99.9%	99.0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	124.1	99.8%	98.2%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	104.9	95.8%	95.0%	Acrodysostosis 2, with or without hormone resistance, 614613
PDHA1	88.0	98.6%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	132.4	100.0%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	134.9	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	106.9	97.6%	88.2%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	115.4	99.3%	95.2%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	126.5	100.0%	99.9%	Prolidase deficiency, 170100
PET100	95.2	99.7%	90.6%	Mitochondrial complex IV deficiency, 220110
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	123.8	100.0%	98.4%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	93.3	100.0%	99.9%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	125.4	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	189.6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	157.0	98.9%	95.7%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	85.8	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	137.4	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.6	100.0%	99.6%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862

PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGAP1	106.2	98.8%	94.3%	Mental retardation, autosomal recessive 42, 615802
PGAP2	145.1	100.0%	99.8%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	74.5	63.7%	60.6%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	47.0	92.1%	78.7%	Phosphoglycerate kinase 1 deficiency, 300653
PGM3	148.4	100.0%	99.9%	Immunodeficiency 23, 615816
PHACTR1	111.1	100.0%	99.8%	Epileptic encephalopathy, early infantile, 70, 618298
PHF21A	97.3	100.0%	99.5%	No OMIM Disease ID
PHF6	57.7	96.9%	84.1%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	78.0	99.5%	96.1%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	126.6	99.4%	96.6%	Chung-Jansen syndrome, 617991
PI4KA	95.6	94.4%	90.4%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	94.8	99.1%	93.8%	Epileptic encephalopathy, early infantile, 80, 618580
PIGC	91.5	99.9%	95.9%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	149.1	100.0%	99.8%	Mental retardation, autosomal recessive 53, 616917
PIGL	132.2	100.0%	99.1%	CHIME syndrome, 280000
PIGN	103.7	93.6%	90.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	157.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	88.3	95.6%	86.3%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGT	169.4	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	93.2	99.8%	98.2%	Glycosylphosphatidylinositol biosynthesis defect 21, 618590
PIGV	129.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	144.9	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	89.9	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	122.5	100.0%	99.8%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500

				Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PIK3R2	115.8	95.4%	91.8%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLAA	162.9	99.9%	98.4%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	134.5	100.0%	99.8%	Epileptic encephalopathy, early infantile, 12, 613722
PLK4	145.8	99.9%	98.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	113.6	99.9%	98.2%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	97.2	99.9%	97.2%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLXND1	134.9	99.8%	98.5%	No OMIM Disease ID
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	113.3	99.6%	97.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121.6	100.0%	99.3%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	123.1	100.0%	100.0%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	113.1	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	153.1	100.0%	99.6%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POGZ	129.7	99.5%	99.2%	White-Sutton syndrome, 616364
POLA1	104.7	99.1%	94.7%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLG	124.4	100.0%	99.8%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR2A	176.1	100.0%	100.0%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	119.8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090

POLR3B	132.0	99.9%	98.3%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	123.6	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	225.4	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	144.2	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	137.5	99.6%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	109.7	100.0%	99.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
PORCN	119.6	99.9%	98.9%	Focal dermal hypoplasia, 305600
POU1F1	109.5	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	65.0	90.8%	76.5%	Snijders Blok-Fisher syndrome, 618604
PPM1D	177.2	100.0%	99.8%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	110.7	100.0%	99.6%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	130.2	100.0%	100.0%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	128.6	99.8%	98.1%	No OMIM Disease ID
PPP2CA	166.7	100.0%	100.0%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	138.5	91.6%	91.6%	Mental retardation, autosomal dominant 36, 616362
PPP2R5B	127.3	100.0%	100.0%	No OMIM Disease ID
PPP2R5C	98.9	97.1%	89.4%	No OMIM Disease ID
PPP2R5D	145.7	100.0%	100.0%	Mental retardation, autosomal dominant 35, 616355
PPP3CA	120.5	99.8%	97.5%	Epileptic encephalopathy, infantile or early childhood, 1, 617711 Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265
PPT1	140.2	90.3%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	173.3	100.0%	100.0%	Renpenning syndrome, 309500
PRKAR1A	80.5	98.1%	92.8%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0

PRMT7	128.8	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	88.9	91.8%	83.0%	Hyperprolinemia, type I, 239500
PRPS1	113.2	100.0%	99.9%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PRR12	146.7	100.0%	98.4%	No OMIM Disease ID
PRSS12	144.0	100.0%	99.8%	Mental retardation, autosomal recessive 1, 249500
PRUNE1	120.3	100.0%	99.6%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PSAP	103.3	100.0%	99.5%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	46.2	91.6%	74.2%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMD12	87.5	98.2%	90.2%	Stankiewicz-Isidor syndrome, 617516
PSPH	122.1	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PTCH1	117.3	100.0%	99.2%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCHD1	143.0	100.0%	100.0%	No OMIM disease ID
PTDSS1	116.2	100.0%	100.0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	125.3	99.7%	95.5%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTF1A	138.4	100.0%	99.7%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTRH2	209.0	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	205.4	100.0%	100.0%	No OMIM Disease ID
PTS	103.1	100.0%	98.3%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	178.6	100.0%	99.6%	Verheij syndrome, 615583
PUM1	132.0	100.0%	99.7%	Spinocerebellar ataxia 47, 617931

PURA	233.4	99.9%	98.8%	Mental retardation, autosomal dominant 31, 616158
PUS1	125.7	99.8%	98.1%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	163.9	100.0%	100.0%	Mental retardation, autosomal recessive 55, 617051
PUS7	134.7	100.0%	99.5%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PYCR1	105.0	100.0%	99.0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	129.0	99.7%	97.6%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	137.7	100.0%	100.0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	103.7	99.9%	99.1%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	142.0	100.0%	99.6%	Ververi-Brady syndrome, 617982
RAB11B	229.2	100.0%	100.0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	80.5	99.7%	95.3%	Warburg micro syndrome 3, 614222
RAB23	102.4	100.0%	99.8%	Carpenter syndrome, 201000
RAB27A	123.3	100.0%	99.6%	Griscelli syndrome, type 2, 607624
RAB39B	108.8	100.0%	100.0%	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	123.3	99.4%	98.8%	Warburg micro syndrome 1, 600118
RAB3GAP2	89.9	99.7%	96.1%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RAC1	108.4	99.5%	95.4%	Mental retardation, autosomal dominant 48, 617751
RAC3	132.6	98.1%	95.4%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	80.8	98.0%	93.5%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RAI1	216.5	100.0%	100.0%	Smith-Magenis syndrome, 182290
RALA	123.4	89.9%	82.9%	No OMIM Disease ID
RARB	93.5	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RARS	93.8	93.3%	88.0%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	102.7	100.0%	99.3%	Pontocerebellar hypoplasia, type 6, 611523
RBBP8	117.9	100.0%	99.3%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBFOX1	145.4	90.6%	89.1%	No OMIM Disease ID
RBM10	122.0	99.9%	98.5%	TARP syndrome, 311900
RBM28	132.1	100.0%	100.0%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079

RBPJ	72.1	95.8%	87.2%	Adams-Oliver syndrome 3, 614814
RCBTB1	98.4	99.9%	99.0%	Retinal dystrophy with or without extraocular anomalies, 617175
RECQL4	181.4	100.0%	100.0%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RELN	131.8	100.0%	99.6%	Lissencephaly 2 (Norman-Roberts type), 257320
RERE	84.5	96.9%	94.3%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	137.7	97.6%	97.1%	No OMIM Disease ID
RFT1	106.6	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015
RHEB	35.8	94.1%	71.9%	No OMIM Disease ID
RHOBTB2	206.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 64, 618004
RIT1	142.5	100.0%	100.0%	Noonan syndrome 8, 615355
RLIM	99.8	99.7%	97.7%	Tonne-Kalscheuer syndrome, 300978
RMND1	130.7	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	143.0	100.0%	100.0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	98.0	99.8%	96.3%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	314.2	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	109.5	95.4%	89.8%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	149.1	100.0%	100.0%	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	178.2	99.9%	98.6%	Tenorio syndrome, 616260
RNF13	80.5	91.4%	71.6%	Epileptic encephalopathy, early infantile, 73, 618379
ROGDI	141.6	100.0%	99.9%	Kohlschutter-Tonz syndrome, 226750
ROR2	176.7	100.0%	99.9%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	105.6	97.1%	91.6%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPL10	68.6	98.5%	89.7%	Mental retardation, X-linked, syndromic, 35, 300998
RPS19	81.5	100.0%	98.3%	Diamond-Blackfan anemia 1, 105650
RPS6KA3	84.8	98.2%	91.5%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600

RRM2B	142.6	100.0%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RSPRY1	143.3	100.0%	100.0%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	74.3	99.1%	94.6%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	145.6	99.8%	98.2%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN4IP1	80.2	99.6%	98.2%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	117.3	98.6%	97.3%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	105.3	100.0%	99.5%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	208.1	100.0%	100.0%	Mental retardation, autosomal recessive 61, 617773
SALL1	127.8	99.9%	99.3%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SAMD9	161.7	100.0%	100.0%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	135.4	100.0%	98.7%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS	112.1	100.0%	99.2%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SATB2	115.6	99.9%	98.3%	Glass syndrome, 612313
SBDS	167.5	100.0%	100.0%	Shwachman-Diamond syndrome, 260400
SC5D	149.4	100.0%	99.6%	Lathosterolosis, 607330
SCAMP5	111.9	100.0%	100.0%	No OMIM Disease ID
SCAPER	137.7	97.8%	96.0%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCN1A	120.1	99.9%	99.0%	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208
SCN1B	186.5	100.0%	99.3%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	135.0	99.5%	97.4%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	139.1	99.9%	98.8%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938

SCN8A	162.9	100.0%	99.9%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SCO1	105.2	100.0%	99.6%	Mitochondrial complex IV deficiency, 220110
SCO2	134.9	100.0%	100.0%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCYL1	161.4	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	123.5	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	94.1	85.1%	78.0%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SEMA3E	131.0	100.0%	99.7%	?CHARGE syndrome, 214800
SEPSECS	160.6	100.0%	100.0%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	110.4	100.0%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	59.0	96.8%	89.5%	Mental retardation, autosomal dominant 58, 618106
SETBP1	129.6	99.1%	97.9%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	170.3	99.8%	99.0%	No OMIM Disease ID
SETD1B	192.2	97.5%	96.6%	No OMIM Disease ID
SETD2	139.1	100.0%	99.7%	Luscan-Lumish syndrome, 616831
SETD5	151.2	100.0%	99.7%	Mental retardation, autosomal dominant 23, 615761
SGPL1	133.4	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SGSH	152.5	98.1%	94.9%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK2	152.8	100.0%	99.9%	No OMIM disease ID
SHANK3	143.0	98.1%	93.8%	Phelan-McDermid syndrome, 606232
SHH	165.7	100.0%	100.0%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	136.8	100.0%	99.4%	Noonan syndrome-like with loose anagen hair, 607721
SHROOM4	104.6	99.9%	98.9%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	131.1	99.8%	98.1%	Epileptic encephalopathy, early infantile, 30, 616341
SIL1	138.5	99.4%	96.7%	Marinesco-Sjogren syndrome, 248800
SIN3A	111.6	99.9%	98.7%	Witteveen-Kolk syndrome, 613406

SIX3	240.1	100.0%	100.0%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	149.5	100.0%	99.7%	Shprintzen-Goldberg syndrome, 182212
SLC12A5	121.0	86.3%	84.2%	Epileptic encephalopathy, early infantile, 34, 616645
SLC12A6	120.5	100.0%	100.0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	155.1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	67.9	99.1%	93.5%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	136.7	98.7%	95.1%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC19A3	139.2	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	149.2	100.0%	99.7%	Dicarboxylic aminoaciduria, 222730
SLC1A2	100.8	99.5%	97.2%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	159.5	100.0%	99.8%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	114.2	99.8%	97.0%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	151.7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	152.1	98.4%	94.4%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	138.5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	133.1	99.7%	99.5%	Fontaine progeroid syndrome, 612289
SLC2A1	160.0	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC33A1	135.7	99.8%	97.0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	123.5	100.0%	99.8%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	114.2	100.0%	99.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	63.5	80.4%	77.0%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	209.1	100.0%	99.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC39A14	101.7	99.9%	98.8%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A8	144.7	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC46A1	121.7	100.0%	98.0%	Folate malabsorption, hereditary, 229050
SLC4A4	114.4	99.8%	97.9%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A1	136.0	100.0%	100.0%	Myoclonic-atonic epilepsy, 616421
SLC6A17	162.2	100.0%	100.0%	Mental retardation, autosomal recessive 48, 616269

SLC6A19	139.1	100.0%	100.0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A3	142.7	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A8	58.5	97.6%	87.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	164.3	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	110.7	100.0%	99.8%	Lysinuric protein intolerance, 222700
SLC9A6	104.0	98.5%	93.3%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	87.8	98.1%	92.1%	Intellectual developmental disorder, X-linked 108, 301024
SMAD4	109.9	100.0%	99.9%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA1	90.8	99.4%	95.0%	No OMIM Disease ID
SMARCA2	109.3	97.3%	96.3%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	163.9	100.0%	99.6%	Coffin-Siris syndrome 4, 614609
SMARCB1	192.9	100.0%	100.0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCC2	104.2	99.9%	98.7%	Coffin-Siris syndrome 8, 618362
SMARCD1	116.1	97.4%	91.7%	No OMIM Disease ID
SMARCE1	66.9	94.6%	85.4%	Coffin-Siris syndrome 5, 616938
SMC1A	93.2	99.9%	98.3%	Cornelia de Lange syndrome 2, 300590
SMC3	82.8	96.4%	89.7%	Cornelia de Lange syndrome 3, 610759
SMG9	100.1	100.0%	100.0%	Heart and brain malformation syndrome, 616920
SMOC1	121.2	99.9%	98.4%	Microphthalmia with limb anomalies, 206920
SMPD1	161.8	100.0%	99.6%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPD4	108.3	99.9%	96.9%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	63.5	88.9%	74.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	121.8	100.0%	99.7%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	182.5	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	140.1	100.0%	99.6%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRPB	83.6	99.9%	98.8%	Cerebrocostomandibular syndrome, 117650
SNRPN	98.3	100.0%	98.4%	Prader-Willi syndrome, 176270
SNX14	79.9	99.7%	93.7%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	199.7	98.9%	98.8%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671

SON	134.3	99.2%	95.4%	ZTTK syndrome, 617140
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	98.0	99.7%	97.7%	Noonan syndrome 9, 616559
SOX10	101.7	100.0%	99.8%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX11	225.3	100.0%	100.0%	Coffin-Siris syndrome 9, 615866
SOX2	261.8	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	87.5	99.0%	95.2%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX4	122.0	100.0%	99.9%	Coffin-Siris syndrome 10, 618506
SOX5	92.0	99.8%	96.6%	Lamb-Shaffer syndrome, 616803
SPART	132.9	100.0%	98.4%	Troyer syndrome, 275900
SPAST	94.9	99.7%	96.9%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	142.5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPECC1L	133.2	100.0%	99.7%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPG11	118.8	99.9%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPOCK1	118.5	100.0%	98.9%	No OMIM Disease ID
SPR	159.7	100.0%	100.0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	143.0	99.8%	98.1%	Legius syndrome, 611431
SPTAN1	118.5	99.1%	98.6%	Epileptic encephalopathy, early infantile, 5, 613477
SPTBN2	141.6	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
SRCAP	166.7	100.0%	99.8%	Floating-Harbor syndrome, 136140
SRD5A3	149.2	99.9%	98.5%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SRPX2	64.2	99.6%	94.4%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SSR4	118.5	100.0%	100.0%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	143.4	100.0%	99.8%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	104.4	89.3%	85.5%	Salt and pepper developmental regression syndrome, 609056

STAG1	108.3	99.8%	96.9%	Mental retardation, autosomal dominant 47, 617635
STAG2	72.0	96.8%	86.9%	Mullegama-Klein-Martinez syndrome, 301022
STAMBP	96.0	99.9%	97.6%	Microcephaly-capillary malformation syndrome, 614261
STIL	153.2	100.0%	99.7%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	125.5	100.0%	99.9%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	112.5	100.0%	99.5%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STT3A	125.4	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127.0	100.0%	99.8%	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	173.7	100.0%	100.0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	108.2	96.8%	96.5%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	57.8	91.5%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	104.2	100.0%	99.7%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUMF1	91.7	99.9%	97.6%	Multiple sulfatase deficiency, 272200
SUOX	180.8	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SURF1	89.9	93.5%	89.1%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SUZ12	102.4	95.1%	87.5%	No OMIM Disease ID
SVBP	114.3	100.0%	100.0%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYN1	73.1	93.6%	84.0%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	64.1	95.0%	83.4%	No OMIM Disease ID
SYNGAP1	152.9	98.5%	98.0%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	126.3	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	87.1	99.9%	98.6%	Mental retardation, X-linked 96, 300802
SYT1	153.0	100.0%	99.1%	Baker-Gordon syndrome, 618218
SZT2	146.3	99.6%	99.5%	Epileptic encephalopathy, early infantile, 18, 615476
TAF1	89.0	99.4%	96.2%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	96.3	100.0%	100.0%	Mental retardation, autosomal recessive 60, 617432
TAF2	110.2	99.8%	98.1%	Mental retardation, autosomal recessive 40, 615599
TAF6	138.7	100.0%	99.5%	Alazami-Yuan syndrome, 617126
TANC2	140.3	99.9%	99.3%	No OMIM Disease ID
TANGO2	139.6	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	132.6	99.0%	97.3%	No OMIM Disease ID
TAT	119.7	100.0%	99.9%	Tyrosinemia, type II, 276600

TBC1D20	121.0	97.4%	94.5%	Warburg micro syndrome 4, 615663
TBC1D23	89.8	98.8%	94.7%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	199.9	100.0%	100.0%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBC1D7	99.0	99.9%	99.0%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	145.8	98.8%	95.5%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	117.0	99.3%	95.6%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	97.5	99.2%	94.9%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	61.2	93.8%	78.4%	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
TBP	103.4	100.0%	99.8%	Spinocerebellar ataxia 17, 607136
TBR1	184.5	100.0%	100.0%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	114.2	93.7%	88.3%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TCF20	134.1	100.0%	100.0%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF4	111.6	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	167.6	99.8%	98.4%	No OMIM disease ID
TCN2	157.6	100.0%	100.0%	Transcobalamin II deficiency, 275350
TCTN2	127.0	100.0%	99.0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDP2	175.8	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	147.6	100.0%	100.0%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	139.6	100.0%	99.9%	Mental retardation, autosomal recessive 14, 614020
TELO2	136.2	99.9%	98.7%	You-Hoover-Fong syndrome, 616954
TFAP2A	124.1	100.0%	99.3%	Branchiooculofacial syndrome, 113620
TGDS	84.3	99.3%	95.0%	Catel-Manzke syndrome, 616145

TGFBR1	156.6	97.3%	94.3%	Loeys-Dietz syndrome 1, 609192
TGIF1	150.3	100.0%	100.0%	Holoprosencephaly 4, 142946
TH	106.8	100.0%	99.2%	Segawa syndrome, recessive, 605407
THOC2	80.5	98.6%	92.0%	Mental retardation, X-linked 12/35, 300957
THOC6	253.7	100.0%	100.0%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	146.5	100.0%	99.3%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIMM50	133.8	100.0%	99.4%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	50.3	95.4%	80.0%	Mohr-Tranebjaerg syndrome, 304700
TINF2	190.9	100.0%	100.0%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TKT	124.8	98.7%	98.3%	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	89.8	98.5%	92.8%	Mental retardation, autosomal dominant 57, 618050
TMCO1	82.6	87.9%	87.3%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM165	159.2	99.9%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	92.0	99.9%	96.9%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	112.1	100.0%	99.7%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	114.5	99.9%	98.8%	Joubert syndrome 14, 614424
TMEM240	184.6	100.0%	100.0%	Spinocerebellar ataxia 21, 607454
TMEM5	167.7	99.8%	96.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TMEM70	117.3	99.9%	98.5%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	87.6	99.9%	96.4%	No OMIM disease ID
TMTC3	90.1	99.9%	97.3%	Lissencephaly 8, 617255
TNIK	107.7	99.9%	98.9%	Mental retardation, autosomal recessive 54, 617028
TOE1	153.2	100.0%	100.0%	Pontocerebellar hypoplasia, type 7, 614969
TP53RK	93.0	99.8%	97.7%	Galloway-Mowat syndrome 4, 617730
TPI1	120.2	99.8%	97.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	146.9	100.0%	99.9%	Thyroid dyshormonogenesis 2A, 274500

TPP1	130.2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TPRKB	57.9	81.5%	74.8%	Galloway-Mowat syndrome 5, 617731
TRAF7	178.6	99.9%	98.7%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	128.8	100.0%	100.0%	Seckel syndrome 9, 616777
TRAPPC11	124.3	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC6B	72.6	100.0%	97.2%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	133.0	100.0%	99.8%	Mental retardation, autosomal recessive 13, 613192
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM8	179.9	100.0%	99.9%	No OMIM Disease ID
TRIO	129.9	99.3%	97.5%	Mental retardation, autosomal dominant 44, 617061
TRIP12	132.7	100.0%	99.3%	Mental retardation, autosomal dominant 49, 617752
TRIT1	107.1	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	130.7	99.9%	98.4%	Mental retardation, autosomal recessive 68, 618302
TRMT10A	116.1	100.0%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	100.7	99.2%	95.3%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM3	125.7	99.8%	98.6%	No OMIM Disease ID
TRRAP	145.6	99.7%	99.2%	Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	117.4	99.6%	98.4%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	155.5	100.0%	100.0%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	93.1	99.9%	96.9%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100.3	100.0%	99.1%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	129.0	99.7%	97.9%	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	123.3	100.0%	99.6%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	222.2	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	111.9	100.0%	99.6%	Mental retardation, X-linked 58, 300210

TTC19	84.9	98.8%	86.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	131.5	99.9%	98.9%	Trichohepatoenteric syndrome 1, 222470
TTC8	116.8	99.7%	97.8%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100.1	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	82.5	99.9%	97.8%	Lissencephaly 3, 611603
TUBA8	136.6	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	123.3	98.0%	94.4%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB2A	83.6	99.8%	98.0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	88.3	100.0%	99.9%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	135.6	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	114.3	97.8%	95.9%	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBG1	162.6	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	108.4	97.8%	95.1%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	169.9	100.0%	99.7%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	155.7	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TWIST1	185.6	100.0%	100.0%	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWNK	170.3	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
UBA5	79.3	96.9%	83.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	122.1	99.6%	97.4%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	80.9	98.8%	92.5%	Angelman syndrome, 105830
UBE3B	119.7	100.0%	99.8%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	118.2	99.9%	98.9%	Johanson-Blizzard syndrome, 243800
UBTF	127.3	100.0%	99.8%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	125.9	100.0%	100.0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	109.2	72.0%	69.8%	Leukodystrophy, hypomyelinating, 14, 617899
UNC13A	131.4	99.5%	97.7%	No OMIM Disease ID
UNC80	114.4	100.0%	99.6%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801

UPB1	150.9	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161
UPF3B	60.7	94.1%	84.6%	Mental retardation, X-linked, syndromic 14, 300676
UROC1	143.4	100.0%	99.9%	?Urocanase deficiency, 276880
USP27X	167.4	100.0%	100.0%	Mental retardation, X-linked 105, 300984
USP7	84.8	95.1%	89.6%	No OMIM Disease ID
USP9X	92.5	97.9%	91.6%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
VAMP1	142.3	100.0%	100.0%	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VAMP2	104.1	99.9%	99.2%	No OMIM Disease ID
VARS	142.4	100.0%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VLDLR	145.5	100.0%	100.0%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	125.6	95.5%	93.8%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	135.9	99.4%	97.8%	Cohen syndrome, 216550
VPS37A	61.7	89.1%	74.8%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	117.2	91.3%	89.9%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	126.7	99.9%	98.3%	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	130.0	100.0%	99.3%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	142.4	100.0%	99.2%	Desanto-Shinawi syndrome, 616708
WARS2	142.1	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	85.7	100.0%	98.7%	No OMIM Disease ID
WASHC4	104.0	98.9%	94.9%	?Mental retardation, autosomal recessive 43, 615817
WDFY3	125.5	100.0%	99.3%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	105.7	97.1%	93.6%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR13	125.7	100.0%	99.8%	No OMIM Disease ID
WDR26	97.7	99.9%	98.4%	Skraban-Deardorff syndrome, 617616
WDR37	152.1	100.0%	98.9%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	152.7	100.0%	100.0%	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45	74.7	97.1%	90.6%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	75.6	97.4%	90.3%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	166.4	100.0%	100.0%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	164.4	100.0%	100.0%	Galloway-Mowat syndrome 1, 251300
WDR81	205.5	100.0%	100.0%	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185

WFS1	210.0	100.0%	99.8%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WWOX	122.0	100.0%	100.0%	Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211
XPA	73.5	99.9%	97.6%	Xeroderma pigmentosum, group A, 278700
XRCC4	139.7	100.0%	99.2%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	138.1	100.0%	99.4%	Desbuquois dysplasia 2, 615777
YME1L1	102.3	98.1%	92.4%	?Optic atrophy 11, 617302
YWHAE	117.3	100.0%	100.0%	No OMIM Disease ID
YWHAG	181.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 56, 617665
YY1	146.4	100.0%	99.1%	Gabriele-de Vries syndrome, 617557
ZBTB11	165.5	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	161.1	100.0%	100.0%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZBTB18	185.2	99.9%	99.4%	Mental retardation, autosomal dominant 22, 612337
ZBTB20	199.5	100.0%	100.0%	Primrose syndrome, 259050
ZBTB24	160.7	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	153.9	99.9%	98.4%	Mental retardation, autosomal recessive 56, 617125
ZC4H2	74.3	99.6%	95.6%	Wieacker-Wolff syndrome, 314580
ZDHHC9	50.3	98.4%	89.1%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	145.0	99.7%	98.6%	Mowat-Wilson syndrome, 235730
ZFYVE26	110.8	99.9%	99.0%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	319.7	100.0%	100.0%	Craniosynostosis 6, 616602
ZIC2	190.6	98.4%	96.3%	Holoprosencephaly 5, 609637
ZMIZ1	155.5	99.9%	99.3%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYND11	119.7	100.0%	99.6%	Mental retardation, autosomal dominant 30, 616083
ZNF148	157.2	100.0%	99.9%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	133.2	99.5%	98.5%	No OMIM Disease ID
ZNF335	147.5	100.0%	99.9%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	161.7	99.9%	99.3%	No OMIM Disease ID
ZNF41	89.3	100.0%	99.6%	No OMIM Disease ID
ZNF462	164.9	100.0%	99.8%	Weiss-Kruszka syndrome, 618619
ZNF711	110.7	99.7%	97.4%	Mental retardation, X-linked 97, 300803

ZSWIM6	127.2	97.5%	95.6%	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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors