INTELLECTUAL DISABILITY GENE PANEL DG 2.14 (1158 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS	124.3	100	99.6	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AASS	128.8	99.6	97.4	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	92.7	100	99.5	GABA-transaminase deficiency, 613163
ABCC9	157.9	99.9	99.2	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	76	74.7	68	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	143.6	99.9	98.3	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	209.6	100	99.9	Chanarin-Dorfman syndrome, 275630
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACO2	129.3	95.8	91.8	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	155.3	100	100	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265
ACSL4	104.7	97.5	91.8	Mental retardation, X-linked 63, 300387
ACTB	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	149.4	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACVR1	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
ACY1	132.8	99.9	98.3	Aminoacylase 1 deficiency, 609924
ADAM22	140.5	99.9	98.6	?Epileptic encephalopathy, early infantile, 61, 617933
ADAR	125	100	99.8	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	82.1	99.5	97.3	Mental retardation, autosomal recessive 36, 615286
ADGRG1	149.7	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752

ADK	100.4	99.5	96.1	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	242.8	100	100	Helsmoortel-van der Aa syndrome, 615873
ADSL	183.6	99.2	99.1	Adenylosuccinase deficiency, 103050
AFF2	122.8	99.9	98.9	Mental retardation, X-linked, FRAXE type, 309548
AFF4	110.7	99.4	97.3	CHOPS syndrome, 616368
AFG3L2	121	91.9	84.9	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGO2	128.5	99.1	99.1	No OMIM phenotype {Epithelial ovarian cancer,reduced risk,association with} (Permuth-Wey (2011) Cancer Res 71,3896)
AGPAT2	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594
AGTR2	205.1	100	100	No OMIM phenotype Mental retardation, X-linked (Ylisaukko-oja (2004) Hum Genet 114, 211) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476) ?Mental retardation, pervasive developmental disorder and epilepsy (Takeshita (2012) Brain Dev epub, epub)
AHCY	124.5	100	99.8	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	120.9	98	96.3	Xia-Gibbs syndrome, 615829
AHI1	139.3	99.2	95.1	Joubert syndrome 3, 608629
AIFM1	106.2	100	99.7	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	84.8	97.3	89.7	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	119.1	93.6	86.9	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	79.6	97.8	88.6	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	131.1	100	99.9	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH3A2	125.7	95.3	94.6	Sjogren-Larsson syndrome, 270200
ALDH4A1	116	100	98.6	Hyperprolinemia, type II, 239510
ALDH5A1	87.6	86.4	80.1	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	50.9	53.6	48.8	Congenital disorder of glycosylation, type Ik, 608540
ALG11	139.6	96.7	96	Congenital disorder of glycosylation, type Ip, 613661

ALG12	156.2	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	86.7	98.7	94.1	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG2	115.9	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.4	96	93.3	Congenital disorder of glycosylation, type Ic, 603147
ALG8	126	96.5	95.1	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	124.3	100	99.6	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ALX1	153.2	99.9	98.4	?Frontonasal dysplasia 3, 613456
ALX4	132.7	98.4	92.5	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMER1	96.9	99.8	98.9	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	72.2	99	94	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	135.5	99.9	99.2	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	173.1	100	100	Glycine encephalopathy, 605899
ANK3	155.1	99.1	98.8	?Mental retardation, autosomal recessive, 37, 615493
ANKH	118.6	100	99.7	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	162.5	98	94.7	?Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	96.3	97.4	94.1	KBG syndrome, 148050
ANO10	116.7	98.8	96.5	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	123	98.3	95.7	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S1	111.3	99.9	99.5	MEDNIK syndrome, 609313
AP1S2	65.7	78.6	70.9	Mental retardation, X-linked syndromic 5, 304340
AP3B1	95	97.8	90.2	Hermansky-Pudlak syndrome 2, 608233
AP3B2	135.1	97.5	94.2	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	121	98.1	97.8	?Hermansky-Pudlak syndrome 10, 617050

AP4B1	147.4	100	99.8	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	98.7	99.7	97.9	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	127.2	99.1	96.4	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	65.8	71.8	69.3	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	96.8	99.7	96.6	Spastic paraplegia 48, autosomal recessive, 613647
APC2	63.5	93.3	85.3	?Sotos syndrome 3, 617169
APOPT1	63.8	81.4	78.1	Mitochondrial complex IV deficiency, 220110
APTX	118.9	94.2	91.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	154.9	100	99.3	Periventricular heterotopia with microcephaly, 608097
ARG1	167.8	100	100	Argininemia, 207800
ARHGAP31	133.4	99.8	98.3	Adams-Oliver syndrome 1, 100300
ARHGEF6	133.7	99.1	95.4	?Mental retardation, X-linked 46, 300436
ARHGEF9	60.2	76.4	74.4	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	150	92.2	89.7	Coffin-Siris syndrome 2, 614607
ARID1B	156.7	94.3	89.5	Coffin-Siris syndrome 1, 135900
ARID2	216.9	99.2	95.7	Coffin-Siris syndrome 6, 617808
ARL13B	97.3	98.9	92.8	Joubert syndrome 8, 612291
ARL6	85.2	99.8	95.3	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARSA	97.8	100	99.7	Metachromatic leukodystrophy, 250100
ARSE	102.1	99.2	93	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	29.1	75.8	59.5	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	105.9	97.6	92.1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASCL1	213.1	90.2	79.5	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASH1L	160.1	98.7	98.5	Mental retardation, autosomal dominant 52, 617796

ASL	114.4	99.9	98.8	Argininosuccinic aciduria, 207900
ASNS	105.8	97.8	90.5	Asparagine synthetase deficiency, 615574
ASPA	127.6	99.1	95.8	Canavan disease, 271900
ASPM	101.2	98	92.2	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	97.9	95.7	87.5	Citrullinemia, 215700
ASXL1	159.8	99.1	97.7	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	152	98.1	98	Shashi-Pena syndrome, 617190
ASXL3	162.9	99.8	98.9	Bainbridge-Ropers syndrome, 615485
ATCAY	146.1	100	99.7	Ataxia, cerebellar, Cayman type, 601238
ATIC	119.5	99.7	99	AICA-ribosiduria due to ATIC deficiency, 608688
ATN1	120.9	99.7	97.7	Dentatorubro-pallidoluysian atrophy, 125370
ATP1A2	190.8	100	99.6	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP2A2	175.2	100	99.9	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP2	46.1	81.2	55.6	?Parkinsonism with spasticity, X-linked, 300911 Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	130	100	99.3	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V1A	144.8	98.2	94.9	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1B2	137	99.9	98.2	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455
ATP7A	133.2	99.7	97.8	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	133.5	100	99.5	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATPAF2	101.5	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	138.3	99.4	96.9	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
ATRX	82.6	98.2	92.2	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040

				Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	90.9	99.9	97.6	3-methylglutaconic aciduria, type I, 250950
AUTS2	110.3	96.9	95.5	Mental retardation, autosomal dominant 26, 615834
AVPR2	127.5	99.3	97.1	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B3GALNT2	115	92.4	89.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	47.5	76.4	71.7	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GLCT	101.2	97.4	93.4	Peters-plus syndrome, 261540
B4GALNT1	151	95.6	90.1	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	105.4	99.9	99	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	104.3	96.1	95	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	120.4	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BBS1	148.9	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.6	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.6	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.8	100	99.8	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.9	99.7	97.3	Bardet-Biedl syndrome 4, 615982
BBS5	81.1	95.8	84.1	Bardet-Biedl syndrome 5, 615983
BBS7	120.7	98.1	91.7	Bardet-Biedl syndrome 7, 615984
BBS9	112.9	96	93.8	Bardet-Biedl syndrome 9, 615986
BCAP31	70.6	93.1	82.5	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	171.5	100	99.5	Maple syrup urine disease, type Ia, 248600
BCKDHB	112.6	88.9	81.3	Maple syrup urine disease, type Ib, 248600
BCL11A	139.3	98.2	97	Dias-Logan syndrome, 617101
BCL11B	79.7	96.6	88.6	?Immunodeficiency 49, 617237
BCOR	109.7	99.3	96.8	Microphthalmia, syndromic 2, 300166
BCORL1	137.6	99.8	98.2	No OMIM phenotype ?Autism (Sanders (2012) Nature 485, 237) Autism spectrum disorder (Jiang (2013) Am J Hum Genet 93, 249) Intellectual disability, coarse face & hypotonia (Schuurs-Hoeijmakers (2013) J Med Genet 50, 802)
BCS1L	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358

				Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	116.3	99.4	96.5	Bloom syndrome, 210900
BRAF	74.4	87.6	77.2	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRAT1	108.5	99.8	97.4	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRD4	81.7	87.5	77	No OMIM phenotype Cornelia de Lange syndrome (Olley (2018) Nat Genet 50, 329
BRF1	100	96.6	92.9	Cerebellofaciodental syndrome, 616202
BRPF1	179.1	100	99.5	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRWD3	97.1	97	92.3	Mental retardation, X-linked 93, 300659
BSCL2	113.5	100	100	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BTD	166.6	100	99.9	Biotinidase deficiency, 253260
BUB1B	136.5	98.6	97.9	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf4	131.3	99.1	94.7	No OMIM phenotype Intellectual disability, autosomal recessive (Philips (2017) Clin Genet 91,100) Intellectual disability, ADHD and hypotonia (Alazami (2015) Cell Rep 10. 148)
C12orf57	152	100	100	Temtamy syndrome, 218340
C12orf65	88.2	97.3	91.9	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C2CD3	143.1	95.8	95.6	?Orofaciodigital syndrome XIV, 615948
C5orf42	122.8	98.6	95.5	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170

CA2	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	124.1	99.5	94.9	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	114.6	96.8	93	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	87.8	92.7	89.1	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1C	154.6	99.9	99.2	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA2D1	82.6	93.1	84.4	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077) Histiocytoid Cardiomyopathy (Cataldo (2014) Cardiol Young epub) West syndrome (Hino-Fukuyo (2015) Hum Genet 134,649)
CACNG2	114.9	100	99.9	?Mental retardation, autosomal dominant 10, 614256
CAD	158.9	100	99.7	Epileptic encephalopathy, early infantile, 50, 616457
CAMK2A	123.2	100	99.9	Mental retardation, autosomal dominant 53, 617798
CAMK2B	103.4	98.7	93.7	Mental retardation, autosomal dominant 54, 617799
CAMTA1	185.9	99.6	98.8	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN10	95.9	99.9	98	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CARS2	121.1	100	99.8	Combined oxidative phosphorylation deficiency 27, 616672
CASK	92.3	98.7	93.7	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CBL	129.8	96.9	95.7	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	116.2	97.1	91.1	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	119.4	99.8	98.3	Mental retardation, autosomal recessive 3, 608443
CC2D2A	127.4	99.5	97.1	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	75.9	98.9	95.5	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510

CCDC115	59.3	88	85.3	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	133.1	98.1	93.3	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	93.3	97.3	89.4	Ritscher-Schinzel syndrome 2, 300963
CCDC78	114.9	100	100	?Centronuclear myopathy 4, 614807
CCDC88A	78.9	94.7	84.9	?PEHO syndrome-like, 617507
CCDC88C	101.4	99.8	97.4	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCND2	152.3	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	82.4	88	76.6	No OMIM phenotype
CDC42	97	96.7	89.5	Takenouchi-Kosaki syndrome, 616737
CDH15	120.6	99.8	97.2	Mental retardation, autosomal dominant 3, 612580
CDK10	114.6	100	99.9	Al Kaissi syndrome, 617694
CDK13	136.6	95.4	88.1	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK5RAP2	123.9	99.9	98.7	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	114.4	94.9	91.8	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1C	21.1	68.1	51.8	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDON	143.8	100	99.6	Holoprosencephaly 11, 614226
CENPF	139.5	99.5	97.6	Stromme syndrome, 243605
CENPJ	141.7	99.7	97.8	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	119.9	99	97.9	Joubert syndrome 25, 616781
CEP120	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	79.2	98.1	89.1	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	162.5	97.2	94.5	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	66.1	88.4	76.7	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	83.5	97.7	89.6	Joubert syndrome 15, 614464
CEP89	125.5	94.7	91.4	No OMIM phenotype

				Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138) ?Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679)
CHAMP1	160.6	100	100	Mental retardation, autosomal dominant 40, 616579
CHD1	95.4	93.7	83.5	Pilarowski-Bjornsson syndrome, 617682
CHD2	137.7	99.3	98.5	Epileptic encephalopathy, childhood-onset, 615369
CHD3	106.1	94.7	92.1	No OMIM phenotype ?Autism (O'Roak (2012) Nature 485,246)
CHD4	131.3	100	99.8	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	148.1	100	99.9	{Autism, susceptibility to, 18}, 615032
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA4	142.1	96.7	95.8	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CIC	54.2	63.7	60.9	Mental retardation, autosomal dominant 45, 617600
CIT	108.6	99.9	98.2	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	161.3	98.9	96.6	Filippi syndrome, 272440
CLCN4	123	100	99.8	Mental retardation, X-linked 49/15, 300114
CLIC2	72.5	99.3	95.2	?Mental retardation, X-linked, syndromic 32, 300886
CLIP1	131.7	99.7	97.8	No OMIM phenotype Intellectual disability, autosomal recessive (Larti (2015) Eur J Hum Genet 23,331)
CLN3	114.9	92.5	90.7	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.1	98.2	92.2	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.6	98.9	95.3	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	163.9	83.5	83.5	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	182.4	100	99.8	Pontocerebellar hypoplasia, type 10, 615803
CLPB	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLTC	171.5	99.8	99.2	Mental retardation, autosomal dominant 56, 617854
CNKSR2	98.1	96.5	89.7	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	188.4	100	99.2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNPY3	87.1	99.8	96.7	Epileptic encephalopathy, early infantile, 60, 617929

CNTNAP2	148	100	99.9	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COASY	168.5	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COG1	124.2	100	99.9	Congenital disorder of glycosylation, type IIg, 611209
COG4	123.8	100	99.9	Congenital disorder of glycosylation, type IIj, 613489
COG5	107	97.4	93.8	Congenital disorder of glycosylation, type IIi, 613612
COG6	78.4	95	85.9	Congenital disorder of glycosylation, type IIl, 614576 Shaheen syndrome, 615328
COG7	125.1	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	122.4	99.9	98.4	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	92.8	97.9	94	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	96.8	98.5	93.9	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	121.6	98.3	92.7	Mental retardation, autosomal dominant 34, 616351
COLEC11	203	100	100	3MC syndrome 2, 265050
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88.4	84.9	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	134.3	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	241.9	100	99.6	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	159.6	100	100	Mitochondrial complex IV deficiency, 220110
CPLX1	79.8	99.9	97.9	Epileptic encephalopathy, early infantile, 63, 617976
CPS1	143.8	100	99.8	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371

				{Venoocclusive disease after bone marrow transplantation}, 0
CRADD	115.2	99.9	98.5	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	130.7	87.8	83.8	Mental retardation, autosomal recessive 2, 607417
CREBBP	123.5	99.4	96.7	Rubinstein-Taybi syndrome 1, 180849
CRLF1	105.6	90.9	89.2	Cold-induced sweating syndrome 1, 272430
CSNK2A1	126.4	94.1	86.2	Okur-Chung neurodevelopmental syndrome, 617062
CSPP1	112	99.8	97.8	Joubert syndrome 21, 615636
CSTB	82.5	97.1	82.7	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	93.4	96.1	85	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTCF	158.4	98.6	96.7	Mental retardation, autosomal dominant 21, 615502
CTDP1	105	86.6	83.6	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNB1	163.8	100	99.9	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNND1	153.8	100	99.9	Blepharocheilodontic syndrome 2, 617681
CTNND2	113.7	93.2	91	No OMIM phenotype Autism (Turner (2015) Nature 520,51) Intellectual disability (Hofmeister (2015) J Med Genet 52,111)
CTSA	134.1	100	99.4	Galactosialidosis, 256540
CTSD	163.7	98	95.3	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	140.7	99.6	97.3	No OMIM phenotype ?Autism (Iossifov (2012) Neuron 74,285)
CUBN	127.8	99.8	98.4	Megaloblastic anemia-1, Finnish type, 261100
CUL4B	72.8	98	88.5	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX1	109.2	94.6	91.9	No OMIM phenotype ?Autism spectrum disorder with intellectual disability (Doan (2016) Cell 167,341)
CUX2	85.6	99.8	97.9	No OMIM phenotype
CWF19L1	119.1	99.5	96.7	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXorf56	91.8	99.7	95.5	?Mental retardation, X-linked 107, 301013
CYB5R3	147.3	98	98	Methemoglobinemia, type I, 250800

				Methemoglobinemia, type II, 250800
CYP27A1	175.1	98.3	96.1	Cerebrotendinous xanthomatosis, 213700
CYP2U1	119.2	93.7	91.2	Spastic paraplegia 56, autosomal recessive, 615030
D2HGDH	134.5	97.5	95.2	D-2-hydroxyglutaric aciduria, 600721
DAG1	220.8	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS2	122.3	100	99.6	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	102.1	97.3	93.8	Maple syrup urine disease, type II, 248600
DCAF17	91.9	95.6	89.3	Woodhouse-Sakati syndrome, 241080
DCC	138.5	100	99.9	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCHS1	160.1	99.8	99.2	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	143.8	100	99.8	Al-Raqad syndrome, 616459
DCX	113.2	100	99.7	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067
DDC	101	99.1	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	149.7	99.9	98	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	113.9	86	81	Warsaw breakage syndrome, 613398
DDX3X	80.5	85.9	82.1	Mental retardation, X-linked 102, 300958
DEAF1	125.9	88.3	83.7	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DENND5A	123	99.8	97.9	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	148.3	99.8	99.3	Epilepsy, familial focal, with variable foci 1, 604364
DHCR24	183	100	100	Desmosterolosis, 602398
DHCR7	158.3	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	93.5	97.8	94.8	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHFR	48.4	91.1	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	141	99.6	98.2	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025

DHX30	160.6	99.9	99.2	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DIAPH1	120.7	99.3	97.8	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	160.3	99.2	98.1	Mental retardation, FRA12A type, 136630
DKC1	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
DLD	123.5	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	90	99.3	94.3	Mental retardation, X-linked 90, 300850
DLG4	170.4	100	99.4	no OMIM phenotype Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394)
DMD	111.5	99.4	97.4	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	117.7	99.9	97.9	Myotonic dystrophy 1, 160900
DNAJC12	129.3	87.4	87.3	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	97.9	98.5	90	3-methylglutaconic aciduria, type V, 610198
DNM1	156.7	89.3	87.5	Epileptic encephalopathy, early infantile, 31, 616346
DNMT3A	115.5	98.8	95.8	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	124.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK6	119.9	98.9	96.5	Adams-Oliver syndrome 2, 614219
DOCK7	114.4	97.9	95.6	Epileptic encephalopathy, early infantile, 23, 615859
DOLK	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768
DONSON	104.9	83.9	78.3	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	102.4	100	97.9	Coffin-Siris syndrome 7, 618027
DPH1	157.1	100	99.7	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DPP6	145.5	96.5	94.5	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPYD	158.3	95.6	93.7	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	133.5	100	99.5	Dihydropyrimidinuria, 222748

DYM	101.3	97.2	94.8	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	179.8	100	99.6	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	159.6	100	100	Mental retardation, autosomal dominant 7, 614104
EBF3	130.4	100	99.1	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	83.3	100	98	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	112.8	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDC3	144.7	100	99.6	?Mental retardation, autosomal recessive 50, 616460
EEF1A2	177.7	98.8	93.8	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFNB2	179.7	99.8	97.7	No OMIM phenotype Anorectal malformation, hypoplastic left ventricle and mild developmental delay (Levy (2018) Clin Genet 93,1141)
EFTUD2	124.2	100	99.4	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	141.4	94	92.9	Kleefstra syndrome 1, 610253
EIF2AK3	147.1	95.1	91.3	Wolcott-Rallison syndrome, 226980
EIF2S3	84.9	97	89.8	MEHMO syndrome, 300148
EIF4A3	106.8	100	99.9	Robin sequence with cleft mandible and limb anomalies, 268305
EIF4G1	135	100	99.5	{Parkinson disease 18}, 614251
ELAC2	123.8	100	99.3	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELOVL4	91.9	99.9	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELP2	125.5	99.2	96.9	Mental retardation, autosomal recessive 58, 617270
EMC1	124.3	100	99.8	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMX2	118	100	100	Schizencephaly, 269160
ENTPD1	165.1	100	99.4	Spastic paraplegia 64, autosomal recessive, 615683
EP300	199.7	99.6	97.9	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPB41L1	128.7	99.9	97.7	?Mental retardation, autosomal dominant 11, 614257

EPG5	126	99.3	97.7	Vici syndrome, 242840
ERCC2	123.7	100	99.7	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.2	99.9	98.9	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC5	139.8	100	99.4	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	191.3	100	99.9	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC8	89.5	92.9	78.4	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	156	100	99.3	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	105.2	97.3	90.4	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	126.6	100	100	Glutaric acidemia IIB, 231680
ETHE1	85.5	99.3	95.8	Ethylmalonic encephalopathy, 602473
EXOC8	174.4	100	100	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
EXOSC2	142	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	88.5	97.3	89.4	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC9	129.8	97.5	87.4	Pontocerebellar hypoplasia, type 1D, 618065
EZH2	139.5	99.8	97.6	Weaver syndrome, 277590
FA2H	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	125.2	97.3	95.2	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	80.4	96.3	92.4	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARSB	76.6	96	93.4	?Neurodevelopmental disorder with brain, liver, and lung abnormalities, 618007
FAT4	224.5	100	99.9	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006

				Van Maldergem syndrome 2, 615546
FBXL4	189.8	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	75.9	93.7	84.9	No OMIM phenotype
FBXO31	108.8	93.5	89	?Mental retardation, autosomal recessive 45, 615979
FGD1	85.7	92.7	86.5	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	95.5	99.6	96.3	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	190.1	100	99.7	Spinocerebellar ataxia 27, 609307
FGFR1	148	99.7	98.3	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	140.1	97.4	96.4	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	110.2	99.6	97	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247

				Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FH	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	123.3	100	100	Thauvin-Robinet-Faivre syndrome, 617107
FIGN	169.6	100	99.9	No OMIM phenotype
FKRP	94.5	100	99.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	120.2	99.2	94.2	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNA	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLVCR1	139.5	99.2	95.8	Ataxia, posterior column, with retinitis pigmentosa, 609033
FMN2	86.6	83.1	77.7	Mental retardation, autosomal recessive 47, 616193
FMR1	78.9	94	84.7	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360

FOXG1	157.5	84.8	81	Rett syndrome, congenital variant, 613454
FOXP1	129.6	100	99.9	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	160.2	98.9	96.9	Speech-language disorder-1, 602081
FRAS1	147.8	100	99.7	Fraser syndrome 1, 219000
FREM2	182.4	100	99.5	Fraser syndrome 2, 617666
FRMD4A	116.7	91.4	90.3	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMPD4	114.1	99.7	98	Mental retardation, X-linked 104, 300983
FRRS1L	103.1	68.3	63.4	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	89.8	94.6	89.8	Glutamate formiminotransferase deficiency, 229100
FTO	118.9	83.7	82.5	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	128.3	98.1	94.5	Mental retardation, X-linked 9/44, 309549
FUCA1	135	100	99.5	Fucosidosis, 230000
FUT8	166.1	99.9	98.8	Congenital disorder of glycosylation with defective fucosylation, 618005
GABBR2	136	95.2	92.4	Epileptic encephalopathy, early infantile, 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, protection against}, 188890 {Nicotine dependence, susceptibility to}, 188890
GABRA1	179.5	100	100	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRA3	110.3	99.6	97.7	No OMIM phenotype Seizures and intellectual disability (Niturad (2017) Brain 140,2879)
GABRB1	174.1	100	99.9	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	148.9	100	100	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	140.5	98.1	93.7	Epileptic encephalopathy, early infantile, 43, 617113 {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GAD1	128.7	99.9	98.4	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	100.6	98.9	94.6	Krabbe disease, 245200
GALE	154.8	100	100	Galactose epimerase deficiency, 230350
GALT	168.7	100	100	Galactosemia, 230400
GAMT	93.5	90.9	80.7	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	129.8	100	99.8	Mental retardation, autosomal dominant 18, 615074
GATM	150.6	100	100	Cerebral creatine deficiency syndrome 3, 612718

GCDH	147.6	99.9	99.1	Glutaricaciduria, type I, 231670
GCH1	74.4	97	86.5	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	34.2	83.1	67.8	?Glycine encephalopathy, 605899
GDI1	152	98.9	97.3	Mental retardation, X-linked 41, 300849
GFAP	102.6	91.7	90.3	Alexander disease, 203450
GFM2	118.6	98.7	93.4	No OMIM phenotype Leigh syndrome with arthrogryposis multiplex congenita (Fukumura (2015) J Hum Genet 60,509) Wolcott-Rallison syndrome (Dixon-Salazar (2012) Sci Transl Med 4,138ra78) {Atorvastatin sensitivity} (Callegari (2012) PLoS Genet 8,e1002755)
GJA1	246.4	100	100	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB1	229.8	100	99.8	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	41.9	68.9	58.6	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	43.6	74	54.7	Glycerol kinase deficiency, 307030
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	78.9	90.6	82.7	Glycine encephalopathy, 605899
GLI2	138.5	99.4	97.4	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	154.2	100	99.7	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800

GLYCTK	202.6	100	99.5	D-glyceric aciduria, 220120
GM2A	139.6	100	100	GM2-gangliosidosis, AB variant, 272750
GMPPA	136.8	100	99.9	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	228.5	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAO1	167.8	93.8	93.8	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAS	141	98.5	95.9	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNB1	189	100	100	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB5	125.8	99.9	98.3	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	133.6	99.4	96.4	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNS	107.9	96.9	92	Mucopolysaccharidosis type IIID, 252940
GPC3	85.1	98.6	92.6	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	167.2	98.4	96.9	Molybdenum cofactor deficiency C, 615501
GPT2	138	98.2	90.9	Mental retardation, autosomal recessive 49, 616281
GRIA3	98.2	99.3	94.7	Mental retardation, X-linked 94, 300699
GRIA4	148.3	99.8	98.1	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	175.4	100	99.9	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	133.2	96	94.7	Mental retardation, autosomal recessive, 6, 611092
GRIN1	150.7	100	99.5	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254

				Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2A	159.1	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	189.4	99.9	99.3	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	73.1	69.1	62.1	Epileptic encephalopathy, early infantile, 46, 617162
GRIN3B	118.3	82	75.1	No OMIM phenotype {Schizophrenia, increased risk,association with} (Matsuno (2015) PLoS One 10,e0116319)
GRIP1	130.8	100	99.9	Fraser syndrome 3, 617667
GRM1	185.8	100	99.9	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRN	184.5	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GSE1	101.5	99.9	98.8	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237)
GSS	104	100	99.8	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	113.6	100	99.1	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	156.8	96.5	94.7	Jaberi-Elahi syndrome, 617988
GTPBP3	137.4	100	99.7	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	116.1	92.2	89.4	Mucopolysaccharidosis VII, 253220
HACE1	125.3	99.2	95	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	106.6	99.9	99.2	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	105.8	99.3	96.1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	122.4	99.9	97.8	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	111.9	99.9	99.3	No OMIM phenotype Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706) Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219) ?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HDAC6	118.7	99.7	97	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	131.9	100	99.8	Cornelia de Lange syndrome 5, 300882

HECTD1	172	98.8	96.1	No OMIM phenotype ?Autism spectrum disorder (Wang (2016) Nat Commun 7,13316)
HECW2	133.1	99.9	98.6	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	142.3	81.4	76.1	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	173.7	99.9	99.4	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	114.4	80.9	77.9	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HESX1	57.6	99.2	92.6	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	118.3	93.8	92.2	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	129.7	99.4	94	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	101	86.4	85.7	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIST1H4C	108.2	100	100	No OMIM phenotype
HIVEP2	191.2	100	100	Mental retardation, autosomal dominant 43, 616977
HLCS	172.8	100	100	Holocarboxylase synthetase deficiency, 253270
HMGCL	143.3	100	99.9	HMG-CoA lyase deficiency, 246450
HNMT	135	100	99.7	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPH2	170.8	100	100	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	71.7	86.9	78.4	Au-Kline syndrome, 616580
HNRNPU	126.6	99.3	97.9	Epileptic encephalopathy, early infantile, 54, 617391
HOXA1	165.3	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HPD	137.8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	58.2	96	84.8	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322

HRAS	164.7	99.8	98.1	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HSD17B10	117.1	100	99.2	HSD10 mitochondrial disease, 300438
HSPA9	91.6	91.1	85.9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	96.5	98.3	93.2	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HUWE1	98.4	99.2	97	Mental retardation, X-linked syndromic, Turner type, 300706
IARS	148.8	99.8	98.6	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093
IARS2	131.5	100	99.9	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IDS	111.3	99.6	98.3	Mucopolysaccharidosis II, 309900
IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	73	93.2	82.2	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	113.5	99.6	97.1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFT172	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT81	92.9	88.3	81.2	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	118.3	99.8	97.7	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	122.5	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	52.5	84.6	73.2	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640

IL1RAPL1	116.8	99.7	98	Mental retardation, X-linked 21/34, 300143
IMPA1	71.4	97.1	85.8	Mental retardation, autosomal recessive 59, 617323
INPP5E	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	108.3	100	99.6	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
IQSEC2	61.2	92.1	82.5	Mental retardation, X-linked 1/78, 309530
ISCA2	92	99.7	96.9	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISPD	104.4	95.2	84.8	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGA7	129.6	99.6	97.6	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	161.4	100	99.9	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	114.9	100	100	Isovaleric acidemia, 243500
JAG1	148.4	98.1	97.5	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAM3	158.6	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	144.5	99.8	97.7	No OMIM phenotype ?Rett syndrome (Saez (2016) Genet Med 18,378) ?Congenital heart disease in 22q11.2 deletion syndrome patients (Guo (2015) Am J Hum Genet 97,869) ?Autism spectrum disorder (Saez (2016) Genet Med 18,378) ?Intellectual disability (Saez (2016) Genet Med 18,378)
KALRN	149	100	99.5	{Coronary heart disease, susceptibility to, 5}, 608901
KANK1	166.2	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	172.3	99.9	99.2	Koolen-De Vries syndrome, 610443
KAT6A	169.3	100	99.8	Mental retardation, autosomal dominant 32, 616268
KAT6B	192.3	99.6	98.5	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	141.7	100	100	Lissencephaly 6, with microcephaly, 616212
KCNA2	157.7	100	99.9	Epileptic encephalopathy, early infantile, 32, 616366
KCNA4	153.1	100	100	No OMIM phenotype

				Abnormal striatum, congenital cataract and intellectual disability (Kaya (2016) J Med Genet 53,786)
KCNB1	145	100	99.9	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	199.2	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	144	68.5	59	Spinocerebellar ataxia 13, 605259
KCNH1	185.8	98.7	98.7	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	213.4	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	299.5	100	100	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ6	156.6	100	99.9	Keppen-Lubinsky syndrome, 614098
KCNK9	193.7	100	100	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	85.4	90.1	86.5	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	110.7	98.9	95.5	Seizures, benign neonatal, 2, 121201
KCNQ5	158	96.2	94.2	Mental retardation, autosomal dominant 46, 617601
KCNT1	112	95.3	92.3	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	166.7	95	95	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	129.7	96.2	93.4	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	138	98.8	96.9	No OMIM phenotype Congenital heart disease (Zaidi (2013) Nature 498,220) ?Intellectual disability (Athanasakis (2014) Am J Med Genet A 164, 170)
KDM5C	112.6	97.9	95.1	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	109	93.2	84.3	Kabuki syndrome 2, 300867
KIAA0586	114.7	98.2	92.7	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1109	144.1	99.1	97.2	Alkuraya-Kucinskas syndrome, 617822
KIDINS220	155.4	99.9	99.5	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	83.8	97.2	94.2	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950

KIF14	111.3	98.1	89.9	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	114	99.2	96.1	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1BP	159.7	96.2	96.1	Goldberg-Shprintzen megacolon syndrome, 609460
KIF2A	105.2	97.7	88.7	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	92.4	98.2	93.7	?Mental retardation, X-linked 100, 300923
KIF5C	116.3	99.9	99.1	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	136.3	100	99.7	Mental retardation, autosomal dominant 4, 612581
KLF7	144.3	100	99.5	No OMIM phenotype ?Developmental delay/intellectual disability, neuromuscular and psychiatric symptoms (Powis (2018) Clin Genet 93,1030)
KLHL15	178.1	100	99.9	Mental retardation, X-linked 103, 300982
KMT2A	152.5	99.3	98.6	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2B	120.3	94	91.2	Dystonia 28, childhood-onset, 617284
KMT2C	170.2	91	88.6	Kleefstra syndrome 2, 617768
KMT2D	142.1	99.9	99	Kabuki syndrome 1, 147920
KMT5B	202.3	100	99.6	Mental retardation, autosomal dominant 51, 617788
KNL1	113.7	98.3	95.2	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	112.1	100	99.9	Mental retardation, autosomal recessive 41, 615637
KRAS	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942

				Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
L1CAM	133.3	99.8	98.4	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	129.1	98.4	97	L-2-hydroxyglutaric aciduria, 236792
LAMA1	137.5	100	99.6	Poretti-Boltshauser syndrome, 615960
LAMA2	143.5	99.9	99.5	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMC3	130.5	98.3	96.1	Cortical malformations, occipital, 614115
LAMP2	106.1	92.7	91.2	Danon disease, 300257
LARGE1	143	100	99.6	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	56.3	80.5	63.7	Alazami syndrome, 615071
LAS1L	90.2	99.7	97.7	Wilson-Turner syndrome, 309585
LIAS	133.7	99.5	97.1	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LINGO1	230.8	100	99.8	No OMIM phenotype ?Intellectual disability (Riazuddin (2017) Mol Psychiatry 22,1604)
LINS1	147.8	99.9	98	Mental retardation, autosomal recessive 27, 614340
LMAN2L	127.9	100	99.8	?Mental retardation, autosomal recessive, 52, 616887
LONP1	141.5	97.9	96.4	CODAS syndrome, 600373
LRP2	176.3	100	99.8	Donnai-Barrow syndrome, 222448
LRPPRC	127.3	99.4	97.2	Leigh syndrome, French-Canadian type, 220111
LZTFL1	109.1	99.1	95.3	Bardet-Biedl syndrome 17, 615994
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAB21L2	245.6	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAF	60.2	77.5	72.7	Ayme-Gripp syndrome, 601088

				Cataract 21, multiple types, 610202
MAG	146.5	100	99.5	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	83.4	90.6	81.2	Schaaf-Yang syndrome, 615547
MAGT1	101.8	98.4	95.8	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAN1B1	128.9	100	99.7	Mental retardation, autosomal recessive 15, 614202
MAN2B1	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAOA	113.8	99.9	98.7	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAP1B	142	100	99.5	No OMIM phenotype Periventricular nodular heterotopia (Heinzen (2018) PLoS Genet 14,e1007)
MAP2K1	92.3	99.8	95.6	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	107.9	97.6	89.2	Cardiofaciocutaneous syndrome 4, 615280
MAPRE2	205.7	100	99.5	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	148.6	100	99.6	3MC syndrome 1, 257920
MAT1A	185.4	99.7	97.5	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	196.2	99.9	99.6	Mental retardation, autosomal dominant 1, 156200
MBOAT7	91.8	99.3	94.7	Mental retardation, autosomal recessive 57, 617188
MBTPS2	113.4	99.6	97.6	?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014
MCCC1	151.7	100	99.4	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	131.1	99.9	98.9	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	150.2	98.8	97	Mucolipidosis IV, 252650
MCPH1	148.6	99.9	98.1	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	123.3	98	97.9	Epileptic encephalopathy, early infantile, 51, 617339
MECP2	87.3	99.1	93.1	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750

				Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MECR	108.1	98.8	96.1	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	105.7	98	94.8	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13	167.6	99.9	99.4	No OMIM phenotype Intellectual disability/developmental delay and speech delay/disorder (Snijders Blok (2018) Hum Genet 137, 375)
MED13L	134.6	100	99.6	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	118	95.2	91.7	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	131.2	98.7	96.9	Mental retardation, autosomal recessive 18, 614249
MED25	103.9	99.1	95.7	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	137.7	97.9	93.5	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEIS2	138.2	100	99.9	Cleft palate, cardiac defects, and mental retardation, 600987
METTL23	145	100	100	Mental retardation, autosomal recessive 44, 615942
MFF	93.7	90.4	87.6	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	115.1	98.8	95.9	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	125.1	99.9	98.4	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	157.2	100	99.9	Congenital disorder of glycosylation, type IIa, 212066
MICU1	134.2	96	88.8	Myopathy with extrapyramidal signs, 615673
MID1	164.6	99.8	98.4	Opitz GBBB syndrome, type I, 300000
MID2	141	99.6	97.3	?Mental retardation, X-linked 101, 300928
MKKS	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.5	99.9	98.5	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	103.4	100	99.8	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	75.8	91.2	86.8	Malonyl-CoA decarboxylase deficiency, 248360

MMAA	183.2	100	99.6	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.2	100	99.9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	205.8	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	77	89.3	75	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	87.4	98.4	93	Molybdenum cofactor deficiency A, 252150
MOCS2	139.5	99.6	99.6	Molybdenum cofactor deficiency B, 252160
MOGS	121.6	99.8	99.1	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	111.8	100	99.7	Congenital disorder of glycosylation, type If, 609180
MPDZ	149	98.7	96.6	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPLKIP	72.5	97.1	79.3	Trichothiodystrophy 4, nonphotosensitive, 234050
MRPL3	66.3	91.2	77.9	Combined oxidative phosphorylation deficiency 9, 614582
MRPS22	138.8	95.3	91.8	Combined oxidative phosphorylation deficiency 5, 611719
MSL3	69.8	94.5	82.7	No OMIM phenotype
MTFMT	124.6	99.3	96.2	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	126.1	98.4	97.2	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTOR	140	100	99.8	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTR	140.9	99.8	98.8	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	139.1	100	99.2	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	121.8	99.2	95.1	Methylmalonic aciduria, mut(0) type, 251000
MVK	124.3	92.1	90.4	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYCN	94.1	94.9	84.8	Feingold syndrome 1, 164280
MYH9	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622

				Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	125.3	99.5	97.4	Griscelli syndrome, type 1, 214450
MYT1L	178.3	100	99.7	Mental retardation, autosomal dominant 39, 616521
NAA10	102.4	98.7	96.7	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	86.6	96.5	90.9	Mental retardation, autosomal dominant 50, 617787
NACC1	167.7	100	99.9	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAGA	139.4	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	108.7	92.4	90.4	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	139.5	99.8	97.5	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	106.1	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	120	97.4	97.1	Combined oxidative phosphorylation deficiency 24, 616239
NBEA	128.4	90.8	89.4	No OMIM phenotype Autism, idiopathic (Castermans (2003) J Med Genet 40, 352) ?Schizophrenia (Fromer (2014) Nature 506, 179) ?Obesity, extreme (Mariman (2015) Physiol Genomics 47,225) ?Tetralogy of Fallot (Silversides (2012) PloS Genet 8)
NBN	80.6	99.1	94.6	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NDE1	100.9	100	99.5	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDP	116.8	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	201.4	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA1	166.8	100	99.6	Mitochondrial complex I deficiency, 252010
NDUFA11	86.9	99.5	95.8	Mitochondrial complex I deficiency, 252010
NDUFA12	160.2	100	100	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000

NDUFA2	133.9	100	100	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF3	120.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF5	95.7	98.8	94.5	Mitochondrial complex 1 deficiency, 252010
NDUFS1	132.2	99.8	98.6	Mitochondrial complex I deficiency, 252010
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFS3	142.4	90.7	90.6	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	147.3	100	99.1	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	119.1	99.9	99.4	Mitochondrial complex I deficiency, 252010
NDUFS7	118.4	100	99.7	Leigh syndrome, 256000
NDUFS8	141.4	100	99.9	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	136.7	99.7	97.8	Mitochondrial complex I deficiency, 252010
NDUFV2	69.5	78.7	53.9	Mitochondrial complex I deficiency, 252010
NECAP1	116.9	100	99.9	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	145.4	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NEDD4L	105.2	71.7	70.5	Periventricular nodular heterotopia 7, 617201
NEU1	148.1	99.4	97.1	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEXMIF	139.2	99.9	99	Mental retardation, X-linked 98, 300912
NF1	125.9	92.3	89.3	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFATC1	136.4	99.2	96.7	No OMIM phenotype Tricuspid atresia (Abdul-Sater(2012) PLoS One 7,e49532) Congenital heart disease (Glessner (2014) Circ Res 115,884) ?Biscuspid aortic valve (Bonachea (2014) BMC Med Genomics 7,56) ?Tetralogy of Fallot (Silversides (2012) PLoS Genet 8, e1002843) ?Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087)
NFIA	145.1	100	99.3	Brain malformations with or without urinary tract defects, 613735
NFIX	165.3	97.7	94.9	Marshall-Smith syndrome, 602535

				Sotos syndrome 2, 614753
NGLY1	128	100	99.5	Congenital disorder of deglycosylation, 615273
NHS	127.1	94.3	93.3	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIPBL	116.1	96.5	94.5	Cornelia de Lange syndrome 1, 122470
NKX2-1	52	96.6	83.3	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NLGN3	128.8	100	99	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	193.6	99.4	97.1	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRP3	150.4	100	100	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
NONO	93.8	99.7	96.4	Mental retardation, X-linked, syndromic 34, 300967
NPC1	147.9	99.2	97.8	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	140.7	100	99.9	Niemann-pick disease, type C2, 607625
NPHP1	117.6	98.8	96.4	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NR2F1	201.6	99.9	98.4	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	149.2	100	100	No OMIM phenotype Schizophrenia (Buervenich (2000) Am J Med Genet 96,808) Parkinson disease (Grimes (2006) Mov Disord 21, 906) Neurology, pediatric (Vissers (2017) Genet Med 19, 1055)
NRAS	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400

				Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NRXN1	160.9	96.8	95.7	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	155.2	100	99.9	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSD2	136.9	99.7	97.8	No OMIM phenotype
NSDHL	169.2	99.9	98.2	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	114.7	95.3	92.2	Mental retardation, autosomal recessive 5, 611091
NTRK1	130.6	99.7	97.7	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	170.8	100	100	Epileptic encephalopathy, early infantile, 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUP62	111.6	100	99.9	Striatonigral degeneration, infantile, 271930
NUS1	69.6	62	40.7	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
OAT	89.2	77.7	70.5	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	220.7	100	100	Pseudo-TORCH syndrome 1, 251290
OCRL	122.2	98.8	96.3	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGT	125.9	100	99.4	Mental retardation, X-linked 106, 300997
OPHN1	89	99.1	96.2	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	106.7	99.9	98.9	Meier-Gorlin syndrome 1, 224690
OSGEP	120.5	100	99.6	Galloway-Mowat syndrome 3, 617729
OTC	123.3	99.9	99.4	Ornithine transcarbamylase deficiency, 311250
OTUD6B	123.4	99.9	98.2	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
PACS1	117.4	97.5	95.7	Schuurs-Hoeijmakers syndrome, 615009
PACS2	150.7	98.4	95.9	Epileptic encephalopathy, early infantile, 66, 618067

PAFAH1B1	105.2	89.1	81.4	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	151.7	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	82.8	97.6	91.8	Mental retardation, X-linked 30/47, 300558
PANK2	146.6	99.3	93.1	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	169	100	100	No OMIM phenotype Intellectual disability, sensorineural hearing loss, skeletal defects and primary ovarian failure (Shao (2016) J Biol Chem 291,12432)
PAX1	132.4	87.7	82.4	?Otofaciocervical syndrome 2, 615560
PAX6	119.9	100	99.9	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX8	94.1	100	99.9	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	111.8	99.3	95.2	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	149.3	97.7	94.6	Pyruvate carboxylase deficiency, 266150
PCCA	103.1	96.4	89.3	Propionicacidemia, 606054
PCCB	129.7	98.7	96.5	Propionicacidemia, 606054
PCDH19	224.1	100	99.3	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	94.5	99.9	98.4	no OMIM phenotype ?Developmental disorder (Fitzgerald (2015) Nature 519,223)
PCLO	165.2	99.7	98.5	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	117.6	98.9	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	101.2	92.8	88.3	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDHA1	109.8	98.1	92.1	Pyruvate dehydrogenase E1-alpha deficiency, 312170

PDP1	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	116.7	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	116	99.6	98.5	Prolidase deficiency, 170100
PET100	94.5	88.8	74.8	Mitochondrial complex IV deficiency, 220110
PEX1	115.8	97.7	95.4	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	111.8	96.1	90.1	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	105.7	99.7	98.3	?Peroxisome biogenesis disorder 14B, 614920
PEX12	168.3	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.6	99.8	98.7	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	137	97.1	93.1	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	92.9	99.9	99.2	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147.1	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	76.4	100	99.8	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	98.1	99.1	94.3	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	111.7	99.9	98.3	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	94.5	90.4	86.1	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	113.5	89.6	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGAP1	98.1	94.9	88.6	Mental retardation, autosomal recessive 42, 615802
PGAP2	158.4	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207

PGAP3	72.8	62.5	58	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	54.5	93.3	81.6	Phosphoglycerate kinase 1 deficiency, 300653
PHF6	62.8	92.6	83.7	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	94	99.8	97.7	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	115.6	100	99.8	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	124.5	95.9	91	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991
PI4KA	112.8	93.6	89.4	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIGA	90.5	90.4	81.3	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	129	99.7	96.4	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	167.4	100	99.7	Mental retardation, autosomal recessive 53, 616917
PIGL	121.8	99.9	99.3	CHIME syndrome, 280000
PIGN	111.3	92.6	87.1	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147	100	99.9	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	171.3	98.1	98	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	145.5	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	147.6	100	99.8	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	121.4	100	99.9	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	120.7	99.9	99.1	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3R2	86.2	89.1	86.1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	117.5	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217

				Parkinson disease 14, autosomal recessive, 612953
PLCB1	142.8	100	99.7	Epileptic encephalopathy, early infantile, 12, 613722
PLK4	145.5	99.5	96.3	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	129.2	100	99.4	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLXND1	110.8	96.9	93.1	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6) Truncus arteriosus (Ta-Shma (2013) Am J Med Genet A 161,3115) {Diabetic nephropathy,association with} (McKnight (2009) Hugo J 3,77)
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	120.8	99.4	96.8	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121	99.7	97.8	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	93	99.8	97.7	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	151.4	100	99.5	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	122.1	99.7	98.5	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POC1A	133.8	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POGZ	168.2	99.4	99.2	White-Sutton syndrome, 616364
POLG	114.4	100	99.5	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR3A	137.4	100	99.9	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	146.4	99.9	98.5	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	127.6	99.7	97.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123

POMGNT2	259.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	205.1	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	155.7	99.7	98.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	111.1	98.9	97.5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PORCN	117.7	100	99.3	Focal dermal hypoplasia, 305600
POU1F1	106.3	98.2	94.7	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	30.1	67.7	57.2	No OMIM phenotype ?Intellectual disability (Dheedene (2014) Mol Syndromol 5,32)
PPM1D	166.7	100	99.8	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450
PPOX	96.1	99.7	98.2	Porphyria variegata, 176200
PPP1CB	96.6	99.6	98.4	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	133.4	99.4	98	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP2R1A	134	91.7	91.6	Mental retardation, autosomal dominant 36, 616362
PPP2R5B	111.3	99.6	95.1	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24, 4775)
PPP2R5C	107.6	95.1	88.2	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24,4775)
PPP2R5D	143.2	100	99.8	Mental retardation, autosomal dominant 35, 616355
PPP3CA	123.8	99	92.9	Epileptic encephalopathy, infantile or early childhood, 1, 617711
PPT1	144.5	90	87.3	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	186.1	100	100	Renpenning syndrome, 309500
PRF1	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	90.7	99.1	93.9	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960

				Pigmented nodular adrenocortical disease, primary, 1, 610489
PRMT7	138.7	100	99.8	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	83.8	84.9	82.3	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	149.5	100	100	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRR12	75	94.6	86.2	No OMIM phenotype
PRSS12	153.3	99.9	98.5	Mental retardation, autosomal recessive 1, 249500
PSAP	114.4	99.9	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.2	91.4	75.8	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSEN1	160.7	100	99.9	?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
PSMD12	76.3	98.1	90.9	Stankiewicz-Isidor syndrome, 617516
PSPH	128.9	98.8	95.4	Phosphoserine phosphatase deficiency, 614023
PTCH1	114.6	98.4	95.9	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCHD1	157.2	100	99.8	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	127.2	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	143.2	99.6	96	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309

				PTEN hamartoma tumor syndrome, 0 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTRH2	279.6	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	144	100	100	No OMIM phenotype ?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry)
PTS	107.2	99.6	94.1	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	173.4	99.9	98.3	Verheij syndrome, 615583
PUM1	158	100	99.9	Spinocerebellar ataxia 47, 617931
PURA	121.8	94.5	87.2	Mental retardation, autosomal dominant 31, 616158
PUS1	127.2	98.6	93.9	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	192	100	100	?Mental retardation, autosomal recessive 55, 617051
PUS7	136.5	99.8	98	No OMIM phenotype ?Intellectual disability (Riazuddin (2017) Mol Psychiatry 22,1604)
PYCR1	86.3	99.4	94.3	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	127.6	100	97.6	Leukodystrophy, hypomyelinating, 10, 616420
QARS	166.6	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	92.3	100	99.5	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	165.9	99.9	98.9	Ververi-Brady syndrome, 617982
RAB11B	249.8	100	100	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	82.7	97.1	86.4	Warburg micro syndrome 3, 614222
RAB27A	143.9	100	99.9	Griscelli syndrome, type 2, 607624
RAB39B	113	100	99.7	?Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	124.2	99.4	98.8	Warburg micro syndrome 1, 600118
RAB3GAP2	94.1	98.4	93.9	Martsolf syndrome, 212720

				Warburg micro syndrome 2, 614225
RAB40AL	174.9	100	100	No OMIM phenotype ?Martin-Probst syndrome (Bedoyan (2012) J Med Genet 49, 332)
RAC1	117.9	97.6	92.8	Mental retardation, autosomal dominant 48, 617751
RAD21	78.5	98.8	94.7	Cornelia de Lange syndrome 4, 614701
RAF1	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAI1	146.3	100	99.7	Smith-Magenis syndrome, 182290
RARB	118.7	100	100	Microphthalmia, syndromic 12, 615524
RARS2	107.2	100	99.1	Pontocerebellar hypoplasia, type 6, 611523
RBFOX1	136.7	89.1	87.8	No OMIM phenotype Epilepsy, rolandic (Lal (2013) PLoS One 8, e73323) Mental retardation (Bhalla (2004) J Hum Genet 49, 308 ?Autism spectrum disorder (Griswold (2015) Mol Autism 6, 43) ?Developmental coordination disorder (Mosca (2016) J Med Genet 53,812) ?Developmental delay (Kamien (2014) Am J Med Genet A 164, 1411)
RBM10	112.1	99.4	95.4	TARP syndrome, 311900
RBM28	138.7	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	89.2	94.1	86.4	Adams-Oliver syndrome 3, 614814
RCBTB1	123.7	100	99.7	Retinal dystrophy with or without extraocular anomalies, 617175
RELN	155.6	100	99.8	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
RERE	71.4	94.9	88.2	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	133.3	97.1	94.5	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6) {Psoriasis,association with} (Strange (2010) Nat Genet 42,985) {Colorectal cancer,increased risk,association with} (Webb (2006) Hum Mol Genet 15,3263)
RFT1	108.3	99.8	97.3	Congenital disorder of glycosylation, type In, 612015
RHEB	39.4	88.7	75.1	No OMIM phenotype
RHOBTB2	227.5	100	100	Epileptic encephalopathy, early infantile, 64, 618004
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RLIM	149.6	99.6	97.8	Mental retardation, X-linked 61, 300978
RMND1	137.2	99.8	97.3	Combined oxidative phosphorylation deficiency 11, 614922

RMRP				Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	142.1	100	99.9	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93.2	87.5	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.2	100	99.9	Aicardi-Goutieres syndrome 3, 610329
RNASET2	96.4	91.9	88.3	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	144.8	100	100	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	177.8	100	99.1	Tenorio syndrome, 616260
ROGDI	112.2	97.9	95.3	Kohlschutter-Tonz syndrome, 226750
RORA	129.8	92.1	87.7	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPGRIP1L	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	85.9	99.1	93.1	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPS6KA3	79.3	94.2	83.3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RSPRY1	168.9	100	99.9	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RTEL1	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTTN	129.5	97.2	94.7	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	104.1	98	97.5	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	182.3	100	99.9	Mental retardation, autosomal recessive 61, 617773
SALL1	138.5	99.3	98.4	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SATB2	110.5	98.5	93.4	Glass syndrome, 612313
SBDS	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SC5D	198.4	100	99.2	Lathosterolosis, 607330
SCAPER	135.9	96	93.6	No OMIM phenotype
SCN1A	135.2	99.6	98	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208

				Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	168.3	97.1	96.1	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2A	156.7	99	96.4	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN8A	198.3	100	99.7	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SCO1	109.6	97.9	94.3	Mitochondrial complex IV deficiency, 220110
SCO2	113.1	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCYL1	143.2	98.6	96.3	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDHA	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SEMA3E	142.6	99.9	99	?CHARGE syndrome, 214800
SEPSECS	159.3	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	55.4	89.9	79.4	No OMIM phenotype
SETBP1	151.7	97.6	96.1	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	119.2	98.6	96.6	No OMIM phenotype Schizophrenia (Takata (2014) Neuron 82, 723)
SETD1B	133.3	96.5	94.6	No OMIM phenotype
SETD2	147.8	100	99.6	Luscan-Lumish syndrome, 616831
SETD5	184.5	100	99.7	Mental retardation, autosomal dominant 23, 615761
SF1	79.1	84.1	75.4	No OMIM phenotype
SGSH	129	95.1	93.6	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK2	128.1	100	99.8	{Autism susceptibility 17}, 613436

SHANK3	84.8	81.2	73.5	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	117.5	99	94	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	100.8	99.8	98.2	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	85.6	97	92.4	Epileptic encephalopathy, early infantile, 30, 616341
SIL1	154.4	99.8	98	Marinesco-Sjogren syndrome, 248800
SIN3A	137.9	100	99.4	Witteveen-Kolk syndrome, 613406
SIX3	145.3	100	98.9	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	85.3	96.4	90.8	Shprintzen-Goldberg syndrome, 182212
SLC12A6	141.8	100	99.9	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	164.1	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	60.3	92.8	82.1	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	119.6	96.8	92.9	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	186.4	100	99.9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	174.2	100	99.9	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A2	128	99.9	99.4	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	156.5	98.9	94.9	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A12	150.5	99.8	98.4	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	192.5	98.8	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	108.7	99.5	96.9	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	115.8	98.6	96.3	Fontaine progeroid syndrome, 612289
SLC2A1	190.1	92.9	92.8	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847

SLC33A1	140.9	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	124	99.9	97.7	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	108.8	99.7	96.8	Congenital disorder of glycosylation, type IIm, 300896
SLC35C1	230.2	99.9	98.4	Congenital disorder of glycosylation, type IIc, 266265
SLC39A12	113.6	99	94.5	No OMIM phenotype
SLC39A8	128.5	100	99.7	Congenital disorder of glycosylation, type IIn, 616721
SLC4A4	122.3	99	97.1	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A1	143.6	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A17	189.8	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A3	145.7	100	99.8	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	56.5	89.8	79.1	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	123.9	100	99.9	Lysinuric protein intolerance, 222700
SLC9A6	104.2	97.6	91.3	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	100.5	80	72	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMARCA2	113.8	95.7	93.7	Nicolaides-Baraitser syndrome, 601358
SMARCA4	143.8	100	99.5	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	214.3	100	100	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCC2	105.7	99.3	97.1	No OMIM phenotype ?Ivemark syndrome (Carss (2014) Hum Mol Genet 23,3269) ?Autism (Neale (2012) Nature 485,242)
SMARCE1	73.6	96.5	86.8	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	99.4	99.9	98.8	Cornelia de Lange syndrome 2, 300590

SMC3	81.4	93.8	87.6	Cornelia de Lange syndrome 3, 610759
SMG9	101.8	100	99.9	Heart and brain malformation syndrome, 616920
SMOC1	129.8	99.5	97.3	Microphthalmia with limb anomalies, 206920
SMPD1	123.5	99.6	97.9	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	67.8	88.3	73.9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	133.7	100	99.9	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	153.5	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	139.3	99.2	96.7	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRPB	75.2	99.8	97.4	Cerebrocostomandibular syndrome, 117650
SNRPN	116.8	99.4	95	Prader-Willi syndrome, 176270
SNX14	70.1	95.2	82.9	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	130.7	92.9	85.4	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	159.7	98.4	94.4	ZTTK syndrome, 617140
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	97.1	98.5	92.8	Noonan syndrome 9, 616559
SOX10	65.8	98.2	91.3	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	118.2	99.7	96.8	Mental retardation, autosomal dominant 27, 615866
SOX2	128.8	98.3	93.1	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	37.7	86.4	71.5	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	107.3	99.1	96.2	Lamb-Shaffer syndrome, 616803
SPART	132.4	99.8	98.2	Troyer syndrome, 275900
SPAST	63.8	93.1	81.9	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	132	99.9	99.2	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPECC1L	157.9	100	100	?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPG11	129.2	99.2	96.9	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360

SPOCK1	118.7	100	99.8	No OMIM phenotype Developmental delay and microcephaly (Dhamija (2014) Eur J Med Genet 57,181)
SPRED1	164.3	98.7	96.7	Legius syndrome, 611431
SPTAN1	125.5	99.1	98.6	Epileptic encephalopathy, early infantile, 5, 613477
SPTBN2	118	99.9	99.3	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SRCAP	153.9	99.8	99.1	Floating-Harbor syndrome, 136140
SRD5A3	135.9	100	99.7	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	81.8	100	98.5	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SSR4	89.8	100	98.8	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	144.5	100	99.9	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	121.9	84.4	84.2	Salt and pepper developmental regression syndrome, 609056
STAG1	104.4	98.8	95.5	Mental retardation, autosomal dominant 47, 617635
STAMBP	112.3	99.3	96.5	Microcephaly-capillary malformation syndrome, 614261
STIL	157.2	99.8	98.6	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	116.5	100	99.9	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STT3A	156.2	100	100	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	125.1	99.2	96	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	152.2	100	98.4	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	124.5	96.8	96.8	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	64.9	93.3	82.8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	101.3	99.6	95.4	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUMF1	103.3	98.6	91.1	Multiple sulfatase deficiency, 272200
SUOX	212.6	100	100	Sulfite oxidase deficiency, 272300
SURF1	96.2	88.3	88.3	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYN1	64.2	74	63.2	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	58.8	92.9	85.2	No OMIM phenotype ?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)

SYNE1	136.6	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNGAP1	141.3	98.4	98	Mental retardation, autosomal dominant 5, 612621
SYP	72	99.8	94	Mental retardation, X-linked 96, 300802
SYT1	171.3	99.8	98.6	No OMIM phenotype Dyskinetic movement disorder, motor delay and cognitive impairment (Baker (2015) J Clin Invest 125, 1670) Intellectual disability and facial abnormalities (Cafiero (2015) Eur J Hum Genet 23, 1499)
SYT14	113.5	59.9	53.8	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	149.5	99.5	99.2	Epileptic encephalopathy, early infantile, 18, 615476
TAF1	112.4	99.4	96.6	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	81.8	100	99.9	Mental retardation, autosomal recessive 60, 617432
TAF2	112.8	98.8	94.7	Mental retardation, autosomal recessive 40, 615599
TAF6	130	99.9	98.6	Alazami-Yuan syndrome, 617126
TANC2	159.1	99.9	99.3	No OMIM phenotype
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	143.1	100	100	Tyrosinemia, type II, 276600
TBC1D20	145.7	94.2	94.1	Warburg micro syndrome 4, 615663
TBC1D24	179.2	100	100	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	105.5	99.6	96.6	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	152.9	95.5	92.3	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	128	99.9	98.2	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	86.5	95.7	89.3	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	79.4	91.5	73	Mental retardation, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBP	129.5	100	98.1	Spinocerebellar ataxia 17, 607136

				{Parkinson disease, susceptibility to}, 168600
TBR1	120.8	100	99.1	No OMIM phenotype Intellectual disability (Hamdan (2014) PLoS Genet 10) ?Autism (O'Roak (2012) Science 338,1619) ?Ventriculomegaly (Traylor (2012) Mol Syndromol 3,102)
TCF20	144.3	100	100	No OMIM phenotype Autism spectrum disorder (Babbs (2014) J Med Genet 51,737)
TCF4	128	99.9	99.5	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	150.4	99.6	96.5	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCN2	175.6	100	100	Transcobalamin II deficiency, 275350
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDP2	165.1	99.9	98.8	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	161.1	100	99.9	Spastic paraplegia 49, autosomal recessive, 615031
TECR	94.6	99.9	97.9	Mental retardation, autosomal recessive 14, 614020
TELO2	98.1	97.4	93.7	You-Hoover-Fong syndrome, 616954
TFAP2A	109.3	100	99.3	Branchiooculofacial syndrome, 113620
TGDS	82.4	98.1	88.8	Catel-Manzke syndrome, 616145
TGFBR1	173.4	93.7	93.6	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	193.5	100	99.9	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	138.3	100	100	Holoprosencephaly 4, 142946
TH	68.2	97.6	88.7	Segawa syndrome, recessive, 605407
THOC2	77.9	96.2	86.6	Mental retardation, X-linked 12/35, 300957
THOC6	248.6	100	99.9	Beaulieu-Boycott-Innes syndrome, 613680
THRB	167.4	100	99.5	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	46	94.5	78.8	Mohr-Tranebjaerg syndrome, 304700
TINF2	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130

TKT	114.1	98.7	97.7	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	113	98.8	94.7	Mental retardation, autosomal dominant 57, 618050
TMCO1	78.7	88	86.5	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM165	113.9	99.8	98.1	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.7	99.8	98.3	Joubert syndrome 14, 614424
TMEM240	112.2	99.8	97.4	Spinocerebellar ataxia 21, 607454
TMEM67	72.9	93.3	83.4	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	138.7	94.6	90.3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	100.9	99.9	97.6	{Autism, susceptibility to, X-linked 6}, 300872
TNIK	111.1	99.9	99.3	Mental retardation, autosomal recessive 54, 617028
TOE1	165.1	100	100	Pontocerebellar hypoplasia, type 7, 614969
TP53RK	37.4	91.3	76.5	Galloway-Mowat syndrome 4, 617730
TPI1	103	99.2	96.7	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	134.8	99.9	98.5	Thyroid dyshormonogenesis 2A, 274500
TPP1	146.3	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRKB	56.7	79.3	67.1	Galloway-Mowat syndrome 5, 617731
TRAF7	147.2	98.3	95	No OMIM phenotype ?Developmental delay, congenital anomalies and dysmorphic features (Tokita (2018) Am J Hum Genet 103,154)
TRAIP	141.6	100	100	Seckel syndrome 9, 616777
TRAPPC11	126.2	99.4	96.4	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC6B	61.8	99.4	94.4	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	135	100	99.9	Mental retardation, autosomal recessive 13, 613192
TREX1	242.4	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700

TRIM32	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110
TRIO	134.3	97.9	95.4	Mental retardation, autosomal dominant 44, 617061
TRIP12	139.8	99.5	98.8	Mental retardation, autosomal dominant 49, 617752
TRIP4	113.5	100	98.8	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	119.4	100	99.8	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	108	99.8	97.4	No OMIM phenotype Intellectual disability (Davarniya (2015) PLoS One 10,e0129631)
TRMT10A	135.2	100	99.4	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	104.6	97.8	92.3	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TSC1	128.8	99.8	98.8	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.2	100	99	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	74.2	99	93.6	Pontocerebellar hypoplasia, type 2F, 617026
TSEN54	82.9	95.9	92.9	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSHB	271.7	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	120.7	99.9	98.6	Mental retardation, X-linked 58, 300210
TTC19	92.1	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	124	99.6	98.1	Trichohepatoenteric syndrome 1, 222470
TTC8	106.9	97.9	92	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	104.5	100	99.7	Mental retardation, autosomal recessive 39, 615541
TUBA1A	113.2	99.9	97.8	Lissencephaly 3, 611603
TUBA8	177.1	99.9	99.7	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	158.6	99.3	97.4	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB2B	100	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031

TUBB3	136.1	98.1	96.9	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	121.2	96	95.3	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	164.2	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	130.8	99.1	96.2	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	152.2	99.9	98.9	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	136.4	100	98.3	Mental retardation, autosomal recessive 7, 611093
TWIST1	134.4	96.6	87.2	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
TWNK	178.8	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
UBA5	75.4	94.1	77.1	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	100.5	99.9	96.9	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	89.8	97.8	91.4	Angelman syndrome, 105830
UBE3B	127.8	100	99.9	Kaufman oculocerebrofacial syndrome, 244450
UBR1	128.2	99.2	96	Johanson-Blizzard syndrome, 243800
UBTF	123.6	99.9	99	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UNC13A	140.8	99.1	97.8	No OMIM phenotype
UNC80	133.4	99.9	99.1	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	157.4	100	100	Beta-ureidopropionase deficiency, 613161
UPF3B	47.4	91.2	76.6	Mental retardation, X-linked, syndromic 14, 300676
UQCRQ	131.3	100	99.9	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	132	99.9	99	?Urocanase deficiency, 276880
USP27X	248.7	100	100	Mental retardation, X-linked 105, 300984
USP7	99.4	93.2	88	No OMIM phenotype ?Autism spectrum disorder (Levy (2011) Neuron 70,886)
USP9X	108.2	97.2	91.1	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968

VLDLR	200.9	99.9	99.4	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	144.9	95.3	93.2	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	143.8	98.6	96.8	Cohen syndrome, 216550
VPS37A	73.6	86.6	66.4	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	129.2	91.4	90.4	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	124.8	97.5	94.2	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	141.5	99.8	98.4	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	165.5	99.7	96.7	Desanto-Shinawi syndrome, 616708
WASF1	85.6	100	99.2	No OMIM phenotype ?Intellectual disability, autistic features and seizures (Ito (2018) Am J Hum Genet 103,144)
WASHC4	91.8	95.3	89.6	?Mental retardation, autosomal recessive 43, 615817
WDR13	122.6	99.9	98.9	No OMIM phenotype Intellectual disability,X-linked (Whibley (2010) Am J Hum Genet 87,173)
WDR19	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR26	97.5	98.3	94.6	Skraban-Deardorff syndrome, 617616
WDR45	75	97.4	90.1	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	85.5	95.8	85.6	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	161.5	100	99.7	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	138.9	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	163.3	99.9	99.4	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WFS1	251.4	100	99.7	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WWOX	130.9	100	99.7	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XPA	52.9	98.5	88.9	Xeroderma pigmentosum, group A, 278700

XPNPEP3	134	100	99.2	Nephronophthisis-like nephropathy 1, 613159
XRCC4	103.2	99.7	97.3	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	132.5	90.4	87.1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.3	98.9	94.9	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	95.6	87.8	81.6	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	105.3	97.7	91.9	?Optic atrophy 11, 617302
YWHAE	114.6	99.7	96.1	No OMIM phenotype Develomental delay,facial dysmorphology and growth retardation (Enomoto (2012) Am J Med Genet A 158A) Developmental delay and mild brain structural abnormalities (Bi (2009) Nat Genet 41,168)
YY1	134.8	100	98.6	Gabriele-de Vries syndrome, 617557
ZBTB16	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	222.7	99.7	99	Mental retardation, autosomal dominant 22, 612337
ZBTB20	216.9	100	100	Primrose syndrome, 259050
ZBTB24	178.1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	184.1	99.6	97.2	Mental retardation, autosomal recessive 56, 617125
ZC4H2	78.6	99.8	98.1	Wieacker-Wolff syndrome, 314580
ZDHHC15	89.8	97.9	92.6	?Mental retardation, X-linked 91, 300577
ZDHHC9	55.5	98.4	89.4	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	157	99.8	98.8	Mowat-Wilson syndrome, 235730
ZFYVE26	120.3	99.9	99.4	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	231.1	100	100	Craniosynostosis 6, 616602
ZIC2	122.5	90.5	78.9	Holoprosencephaly 5, 609637
ZMYND11	137	100	99.8	Mental retardation, autosomal dominant 30, 616083
ZNF292	134.7	98.8	97.2	No OMIM phenotype ?Autism (Neale (2012) Nature 485,242)
ZNF407	176.8	99.2	98.4	No OMIM phenotype Intellectual disability and autism (Ren (2013) Biochim Biophys Acta 1832,431) Cognitive impairment, failure to thrive, hypotonia and dysmorfic features (Kambouris (2014) Orphanet J Rare Dis 9)

ZNF41	103.3	100	99.7	Mental retardation, X-linked 89, 300848
ZNF592	150.1	100	99.9	Spinocerebellar ataxia, autosomal recessive 5, 251300
ZNF711	137.7	98.7	95.5	Mental retardation, X-linked 97, 300803
ZNF81	90.3	98.9	95.6	Mental retardation, X-linked 45, 300498
ZSWIM6	150.9	93.1	89.1	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
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 : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors