INTELLECTUAL DISABILITY GENE PANEL DG 2.9 / DG 2.10 (1034 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
A2ML1	148.8	100%	99%	No OMIM phenotype Noonan-like syndrome (Vissers et al. 2015) Noonan syndrome (van Trier (2015) Int J Pediatr Otorhinolaryngol, epub) Otitis media, susceptibility to (Santos-Cortez (2015) Nat Genet 47,917)
AARS	145.1	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AASS	169.7	100%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	103.8	100%	99%	GABA-transaminase deficiency, 613163
ABCC9	175	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	94.7	76%	69%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	152.7	99%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	256.2	100%	99%	Chanarin-Dorfman syndrome, 275630
ACAD9	157.1	99%	97%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACO2	141.2	97%	94%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	167.6	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	146.1	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	128.1	97%	93%	Mental retardation, X-linked 63, 300387
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	129.7	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACVR1	184.8	100%	99%	Fibrodysplasia ossificans progressiva, 135100
ACY1	162.7	99%	98%	Aminoacylase 1 deficiency, 609924
ADAM22	171.2	100%	99%	No OMIM phenotype

				Epilepsy with progressive encephalopathy and cortical atrophy (Muona (2016) Neurol Genet 2) ?Autism (Neale (2012) Nature 485,242)
ADAR	136.8	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	82.8	99%	97%	Mental retardation, autosomal recessive 36, 615286
ADCK3	161	99%	99%	Coenzyme Q10 deficiency, primary, 4, 612016
ADK	118.9	99%	98%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	276.9	100%	100%	Helsmoortel-van der Aa syndrome, 615873
ADSL	196.2	100%	100%	Adenylosuccinase deficiency, 103050
AFF2	145.1	99%	99%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	141.5	99%	98%	CHOPS syndrome, 616368
AFG3L2	124	94%	87%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	175.8	100%	100%	Aspartylglucosaminuria, 208400
AGO2	141.7	99%	99%	No OMIM phenotype {Epithelial ovarian cancer,reduced risk,association with} (Permuth-Wey (2011) Cancer Res 71,3896)
AGPAT2	117.9	99%	95%	Lipodystrophy, congenital generalized, type 1, 608594
AGTR2	246.8	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	132.6	100%	98%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	137.5	99%	97%	Xia-Gibbs syndrome, 615829
AHI1	176.9	99%	98%	Joubert syndrome-3, 608629
AIFM1	126	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	109.1	99%	94%	Leukodystrophy, hypomyelinating, 3, 260600
AKT3	97.6	99%	95%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	143.9	100%	100%	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	153.1	100%	99%	Sjogren-Larsson syndrome, 270200
ALDH4A1	137.2	100%	99%	Hyperprolinemia, type II, 239510
ALDH5A1	105.3	92%	84%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	57.5	53%	49%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	189.4	100%	99%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	171.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	104.2	99%	96%	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	115.3	100%	99%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	116.7	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	126.3	98%	95%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	147.2	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	128.6	99%	99%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	208.7	99%	99%	Alstrom syndrome, 203800
ALX1	174.7	99%	98%	?Frontonasal dysplasia 3, 613456
ALX4	145.9	99%	94%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMMECR1	87.6	98%	94%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMPD2	153.4	100%	99%	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	187.9	100%	100%	Glycine encephalopathy, 605899
ANK3	189.9	99%	99%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	124.8	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	187.9	97%	94%	?Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	118.5	98%	95%	KBG syndrome, 148050
ANO10	136.7	99%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	144.4	98%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S1	114.6	100%	99%	MEDNIK syndrome, 609313
AP1S2	77	81%	68%	Mental retardation, X-linked syndromic 5, 304340
AP3B1	128.1	99%	95%	Hermansky-Pudlak syndrome 2, 608233

AP3B2	153.3	97%	93%	Epileptic encephalopathy, early infantile, 48
AP4B1	163.4	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	125.7	99%	98%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	133.2	99%	97%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	83.2	76%	70%	Spastic paraplegia 52, autosomal recessive, 614067
APC2	64.9	93%	84%	?Sotos syndrome 3, 617169
APTX	127.2	94%	92%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	172.3	99%	99%	Periventricular heterotopia with microcephaly, 608097
ARG1	181	100%	100%	Argininemia, 207800
ARHGAP31	141.8	99%	98%	Adams-Oliver syndrome 1, 100300
ARHGEF6	154.5	97%	93%	Mental retardation, X-linked 46, 300436
ARHGEF9	99.5	99%	98%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	158.4	95%	91%	Coffin-Siris syndrome 2, 614607
ARID1B	159.9	96%	92%	Coffin-Siris syndrome 1, 135900
ARID2	221.7	99%	98%	No OMIM phenotype Intellectual disability (Shang (2015) Neurogenetics epub) ?Schizophrenia (Fromer (2014) Nature 506,179)
ARL13B	117.5	99%	97%	Joubert syndrome 8, 612291
ARL6	121.4	100%	97%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARSA	114.5	100%	99%	Metachromatic leukodystrophy, 250100
ARSE	107.3	98%	93%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	38.3	81%	67%	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	147.1	99%	97%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASCL1	208.2	95%	84%	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880

ASL	128.8	100%	98%	Argininosuccinic aciduria, 207900
ASNS	108.7	98%	94%	Asparagine synthetase deficiency, 615574
ASPA	144.7	99%	94%	Canavan disease, 271900
ASPM	134.5	99%	96%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	118.2	96%	90%	Citrullinemia, 215700
ASXL1	172.5	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	174.3	98%	98%	Shashi-Pena syndrome, 617190
ASXL3	170	99%	99%	Bainbridge-Ropers syndrome, 615485
ATCAY	165.5	100%	99%	Ataxia, cerebellar, Cayman type, 601238
ATIC	142.5	99%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATN1	135.7	99%	97%	Dentatorubro-pallidoluysian atrophy, 125370
ATP1A2	219	100%	99%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP2A2	198.5	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP2	55.5	91%	71%	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	160.2	99%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V1B2	168.1	100%	99%	Deafness,congenital,with onychodystrophy,autosomal dominant, 124480 Zimmerman-Laband syndrome 2, 616455
ATP7A	148.2	99%	98%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	149.7	100%	99%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATPAF2	115.8	100%	99%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	175.1	99%	98%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	105.7	98%	95%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	103	99%	96%	3-methylglutaconic aciduria, type I, 250950

AUTS2	125.8	97%	96%	Mental retardation, autosomal dominant 26,615834
AVPR2	149.9	99%	96%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B3GALNT2	141.5	93%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	56.4	76%	71%	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GALTL	120.7	98%	95%	Peters-plus syndrome, 261540
B3GNT1	125.5	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 13, 615287
B4GALNT1	167.7	99%	96%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	124.3	100%	99%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	122.7	97%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BBS1	178.6	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	195.4	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	237	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	201.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	153.5	99%	98%	Bardet-Biedl syndrome 4, 615982
BBS5	144	98%	96%	Bardet-Biedl syndrome 5, 615983
BBS7	156.1	99%	95%	Bardet-Biedl syndrome 7, 615984
BBS9	139.9	97%	95%	Bardet-Biedl syndrome 9, 615986
BCAP31	75.6	94%	81%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	195.9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	147.9	93%	84%	Maple syrup urine disease, type Ib, 248600
BCL11A	150.4	98%	97%	Intellectual development disorder with persistence of fetal hemoglobin, 617101
BCOR	117.5	99%	97%	Microphthalmia, syndromic 2, 300166
BCORL1	152.6	99%	97%	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	199	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	139.5	99%	97%	Bloom syndrome, 210900

BRAF	86.4	91%	82%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
BRAT1	121.6	99%	98%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRF1	112.5	99%	95%	Cerebellofaciodental syndrome, 616202
BRWD3	122.6	98%	94%	Mental retardation, X-linked 93, 300659
BSCL2	129.8	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	168.1	100%	100%	Biotinidase deficiency, 253260
BUB1B	158.2	98%	98%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf2	192.9	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
C12orf4	141.9	100%	99%	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	150	100%	99%	Temtamy syndrome, 218340
C12orf65	94.5	98%	93%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C2CD3	155.7	95%	95%	?Orofaciodigital syndrome XIV, 615948
C5orf42	154.1	99%	97%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	178.8	99%	97%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	131.6	99%	96%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	131.4	97%	93%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	104.4	95%	92%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500

				Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1C	182.7	99%	99%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA2D1	111.5	96%	90%	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	131.8	100%	99%	Mental retardation, autosomal dominant 10, 614256
CAD	174.8	99%	99%	?Congenital disorder of glycosylation, type Iz, 616457
CAMTA1	203.2	100%	99%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN10	111.7	100%	99%	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CASC5	129.5	98%	96%	Microcephaly 4, primary, autosomal recessive, 604321
CASK	112.2	99%	95%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CBL	146	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	134.1	98%	94%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	141.1	100%	99%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	144.7	99%	97%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	79.9	99%	94%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC174	133.9	99%	95%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	109.1	98%	92%	Ritscher-Schinzel syndrome 2, 300963
CCDC78	120.3	100%	99%	Myopathy, centronuclear, 4, 614807
CCDC88C	119.9	99%	98%	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053
CCND2	169.6	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CDH15	145.3	99%	97%	Mental retardation, autosomal dominant 3, 612580
CDK5RAP2	142.1	99%	99%	Microcephaly 3, primary, autosomal recessive, 604804

CDKL5	135.2	99%	96%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1C	37.4	77%	63%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDON	147.2	100%	99%	Holoprosencephaly 11, 614226
CENPJ	171.4	99%	99%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP135	98.6	98%	91%	?Microcephaly 8, primary, autosomal recessive, 614673
CEP152	204.7	98%	96%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	92.8	95%	87%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	98.2	98%	93%	Joubert syndrome 15, 614464
CEP89	162	99%	98%	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	193.5	100%	100%	Mental retardation, autosomal dominant 40, 616579
CHD2	145.9	99%	99%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	116.7	97%	93%	No OMIM phenotype ?Autism (O'Roak (2012) Nature 485,246)
CHD4	145.7	100%	99%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	168.4	100%	99%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	168.4	100%	99%	{Autism, susceptibility to, 18}, 615032
CHKB	102.6	99%	97%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA4	169.5	97%	96%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CIC	58.8	63%	61%	No OMIM phenotype Intellectual disability (Vissers (2010) Nat Genet 42,1109)
CIT	122.9	99%	98%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	219	99%	97%	Filippi syndrome, 272440
CLCN4	131	100%	99%	Mental retardation, X-linked 49/15, 300114

CLCNKB	113.9	99%	94%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLIC2	84.3	99%	95%	?Mental retardation, X-linked, syndromic 32, 300886
CLIP1	158.8	99%	98%	No OMIM phenotype Intellectual disability, autosomal recessive (Larti (2015) Eur J Hum Genet 23,331)
CLN3	133	99%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	155.4	99%	97%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	144.8	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	273.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	185.2	100%	100%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	153.8	96%	96%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CNKSR2	105.6	96%	89%	No OMIM phenotype Intellectual disability,X-linked non syndromic (Vaags (2014) Ann Neurol 76,758)
CNNM2	228.4	100%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTNAP2	168.7	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COASY	173	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COG1	129.4	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	139.6	100%	99%	Congenital disorder of glycosylation, type IIj, 613489
COG5	135.2	99%	95%	Congenital disorder of glycosylation, type IIi, 613612
COG6	103.3	97%	90%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	139.6	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	118.4	99%	98%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	103.5	98%	94%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	111.5	99%	96%	Porencephaly 2, 614483

				{Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	153.5	99%	98%	Mental retardation, autosomal dominant 34, 616351
COLEC11	221.8	100%	99%	3MC syndrome 2, 265050
COQ2	92.8	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	103.5	87%	84%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ9	94.2	99%	96%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	250.6	100%	99%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	103.8	100%	98%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	186.7	100%	100%	Mitochondrial complex IV deficiency, 220110
CPS1	183.3	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CRADD	131	99%	97%	Mental retardation, autosomal recessive 34, with variant lissencephaly 614499
CRBN	175.6	100%	98%	Mental retardation, autosomal recessive 2, 607417
CREBBP	141.3	98%	96%	Rubinstein-Taybi syndrome, 180849
CRLF1	127.2	91%	90%	Cold-induced sweating syndrome 1, 272430
CSNK2A1	144.7	95%	89%	Okur-Chung neurodevelopmental syndrome, 617062 Glaucoma, primary congenital (Lee (2011) Mol Vis 17,3583)
CSPP1	133.7	99%	98%	Joubert syndrome 21, 615636
CTBP1	109.1	97%	96%	No OMIM phenotype Developmental delay, hypotonia, ataxia and tooth enamel defects (Beck (2016) Neurogenetics epub,epub)
CTCF	166.6	99%	97%	Mental retardation, autosomal dominant 21, 615502
CTDP1	120.5	91%	84%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNB1	199.2	100%	100%	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNND1	165.2	100%	99%	No OMIM phenotype ?Autism (O'Roak (2012) Nature 485,246)

CTNND2	121.9	94%	91%	No OMIM phenotype Autism (Turner (2015) Nature 520,51) Intellectual disability (Hofmeister (2015) J Med Genet 52,111)
CTSA	153.2	99%	99%	Galactosialidosis, 256540
CTSD	197.2	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	163.9	99%	98%	No OMIM phenotype ?Autism (Iossifov (2012) Neuron 74,285)
CUBN	134	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CUL4B	88.6	98%	92%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CWF19L1	139.1	99%	98%	?Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5R3	181.8	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP27A1	206.9	98%	96%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	162.9	95%	93%	Spastic paraplegia 56, autosomal recessive, 615030
D2HGDH	153.2	98%	96%	D-2-hydroxyglutaric aciduria, 600721
DAG1	236.1	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	155.1	100%	99%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	138.6	99%	96%	Maple syrup urine disease, type II, 248600
DCAF17	125.5	99%	97%	Woodhouse-Sakati syndrome, 241080
DCC	160.1	100%	99%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239 Mirror movements 1, 157600
DCHS1	172.3	99%	99%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	174.5	100%	99%	Al-Raqad syndrome, 616459
DCX	123.8	100%	99%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDC	121.6	99%	97%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	175.7	99%	98%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	104.7	79%	74%	Warsaw breakage syndrome, 613398
DDX3X	109.8	98%	95%	Mental retardation, X-linked 102, 300958
DEAF1	147.6	90%	84%	Mental retardation, autosomal dominant 24, 615828
DENND5A	126.6	99%	99%	Epileptic encephalopathy, early infantile, 49, 617281

DEPDC5	164.8	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DHCR24	205.7	100%	100%	Desmosterolosis, 602398
DHCR7	173	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHFR	61.8	95%	81%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	156.7	99%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIAPH1	131.4	99%	98%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	183.3	99%	99%	Mental retardation, FRA12A type, 136630
DKC1	122.5	100%	98%	Dyskeratosis congenita, X-linked, 305000
DLD	157	99%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	102.3	99%	94%	Mental retardation, X-linked 90, 300850
DLG4	184.2	100%	99%	no OMIM phenotype Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394)
DMD	140.4	99%	98%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	132.8	99%	98%	Myotonic dystrophy 1, 160900
DNAJC12	165.1	87%	87%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	115.5	98%	94%	3-methylglutaconic aciduria, type V, 610198
DNM1	167.4	94%	90%	Epileptic encephalopathy, early infantile, 31, 616346
DNMT3A	127.7	99%	96%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	143.6	99%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK6	134.7	99%	97%	Adams-Oliver syndrome 2, 614219
DOCK7	147.1	98%	96%	Epileptic encephalopathy, early infantile, 23, 615859
DOLK	189.5	100%	99%	Congenital disorder of glycosylation, type Im, 610768
DONSON	119.8	92%	85%	No OMIM phenotype Microcephalic dwarfism (Reynolds (2017) Nat Genet 49,537)
DPAGT1	119.1	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	180	100%	99%	Developmental delay with short stature,dysmorphic features and sparse hair,616901
DPM1	149.2	92%	87%	Congenital disorder of glycosylation, type Ie, 608799
DPP6	157.8	98%	95%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956

DPYD	190.3	97%	95%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	144.7	99%	98%	Dihydropyrimidinuria, 222748
DYM	116.7	97%	96%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	189.4	100%	99%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	168	100%	99%	Mental retardation, autosomal dominant 7, 614104
EBP	89.2	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EDC3	147.9	100%	99%	?Mental retardation, autosomal recessive 50, 616460
EEF1A2	213.2	100%	98%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFTUD2	127.2	99%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	163	99%	98%	Kleefstra syndrome, 610253
EIF2AK3	177.3	96%	91%	Wolcott-Rallison syndrome, 226980
EIF4A3	115.1	100%	99%	Robin sequence with cleft mandible and limb abnormalities,268305
EIF4G1	150.3	100%	99%	{Parkinson disease 18}, 614251
ELAC2	133	100%	99%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELOVL4	115.5	100%	99%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
ELP2	145.1	99%	97%	Mental retardation, autosomal recessive 58, 617270
EMC1	133.2	100%	99%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMX2	132.8	100%	100%	Schizencephaly, 269160
ENTPD1	170.4	99%	99%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	204.1	99%	98%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPB41L1	150.6	100%	99%	?Mental retardation, autosomal dominant 11, 614257
EPG5	138.8	99%	98%	Vici syndrome, 242840
ERCC2	143	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675

				Xeroderma pigmentosum, group D, 278730
ERCC3	120.8	100%	99%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC5	151.3	100%	99%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	196.3	100%	99%	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	109.4	96%	87%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	172.1	100%	99%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	135.2	98%	94%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	128.4	100%	100%	Glutaric acidemia IIB, 231680
ETHE1	92	99%	97%	Ethylmalonic encephalopathy, 602473
EXOSC2	146.2	100%	100%	No OMIM phenotype Retinitis pigmentosa, hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt (Di Donato (2016) J Med Genet 53,419)
EXOSC3	90.8	97%	91%	Pontocerebellar hypoplasia, type 1B, 614678
EZH2	166.9	99%	98%	Weaver syndrome, 277590
FA2H	101.9	95%	89%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	161.3	98%	96%	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	102.4	96%	93%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FAT4	259.2	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBXL4	231	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	105.7	94%	90%	?Mental retardation, autosomal recessive 45, 615979
FGD1	98.4	94%	88%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	114.7	99%	96%	?Epileptic encephalopathy, early infantile, 47, 617166

FGF14	225.2	100%	99%	Spinocerebellar ataxia 27, 609307
FGFR1	165.2	99%	98%	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	155.6	97%	96%	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 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 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	129.1	100%	99%	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	183.5	93%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	135.1	100%	100%	Thauvin-Robinet-Faivre syndrome, 617107
FIGN	161.9	100%	99%	No OMIM phenotype
FKRP	103.2	100%	100%	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	155.6	99%	95%	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	160.7	100%	99%	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLVCR1	166.7	99%	98%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FMN2	107.3	84%	79%	Mental retardation, autosomal recessive 47, 616193
FMR1	90.5	97%	90%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FOXG1	139.8	87%	81%	Rett syndrome, congenital variant, 613454
FOXP1	155.1	100%	100%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	184.8	99%	98%	Speech-language disorder-1, 602081
FRAS1	168.5	100%	99%	Fraser syndrome, 219000
FREM2	194.3	99%	99%	Fraser syndrome, 219000
FRMD4A	129.8	92%	90%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819

FRMPD4	124.4	99%	97%	Mental retardation,X-linked 104, 300983
FRRS1L	130.4	70%	62%	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	104.6	95%	89%	Glutamate formiminotransferase deficiency, 229100
FTO	148.3	100%	99%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	143.1	99%	96%	Mental retardation, X-linked 9, 309549
FUCA1	151.4	100%	99%	Fucosidosis, 230000
GABRA1	219.1	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB1	207.2	100%	100%	Epileptic encephalopathy, early infantile, 45
GABRB3	158	97%	93%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269
GAD1	139.7	99%	98%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALE	182.8	100%	99%	Galactose epimerase deficiency, 230350
GALT	182.8	100%	100%	Galactosemia, 230400
GAMT	123	98%	91%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	140.6	100%	99%	Mental retardation, autosomal dominant 18, 615074
GATM	173	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GCDH	139.2	93%	91%	Glutaricaciduria, type I, 231670
GCH1	92.5	97%	89%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	41.7	85%	70%	Glycine encephalopathy, 605899
GDI1	160.8	100%	100%	Mental retardation, X-linked 41, 300849
GFAP	119.9	99%	98%	Alexander disease, 203450
GFM2	154.5	99%	97%	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	238.2	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850

				Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJC2	50	88%	71%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	57.5	84%	68%	Glycerol kinase deficiency, 307030
GLB1	97.3	99%	97%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	88.5	92%	84%	Glycine encephalopathy, 605899
GLI2	148.1	99%	97%	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	168.4	99%	99%	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	230.9	99%	99%	D-glyceric aciduria, 220120
GM2A	154.1	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPA	162.5	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	276.3	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	197.4	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GNAS	154.4	99%	97%	Acromegaly, somatic, 102200 ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNB1	225.2	100%	100%	Leukemia,acute lymphoblastic,somatic, 613065 Mental retardation, autosomal dominant 42, 616973

GNB5	144.8	99%	98%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	176.5	99%	97%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	190.4	99%	97%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNS	119.6	97%	93%	Mucopolysaccharidosis type IIID, 252940
GPC3	106.4	98%	94%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	186.4	99%	97%	Molybdenum cofactor deficiency C, 615501
GPR56	177.1	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
GPT2	160.5	99%	94%	?Mental retardation, autosomal recessive 49, 616281
GRIA3	102.9	99%	95%	Mental retardation, X-linked 94, 300699
GRID2	216.8	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	170.5	96%	95%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	169.2	100%	99%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	170.4	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	194.9	99%	99%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN3B	121.7	87%	80%	No OMIM phenotype {Schizophrenia, increased risk,association with} (Matsuno (2015) PLoS One 10,e0116319)
GRIP1	148.9	100%	99%	Fraser syndrome, 219000
GRM1	194.2	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSE1	102.6	99%	98%	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237)
GSS	117	100%	99%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	152.5	100%	99%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP3	139.5	100%	99%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	126.8	91%	87%	Mucopolysaccharidosis VII, 253220
HACE1	167.6	99%	98%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HAX1	157.7	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	115.6	99%	98%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	113.8	99%	96%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541

HCN1	155.1	100%	99%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	121.1	100%	99%	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	124.6	99%	97%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	142.5	100%	99%	Cornelia de Lange syndrome 5, 300882
HECTD1	193	99%	98%	No OMIM phenotype ?Autism spectrum disorder (Wang (2016) Nat Commun 7,13316)
HECW2	148.8	99%	99%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	156.4	86%	78%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	191.4	99%	99%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	120.8	79%	76%	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	96.7	99%	96%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	133.7	100%	98%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	166.2	99%	95%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HIVEP2	214.5	100%	100%	Mental retardation, autosomal dominant 43, 616977
HLCS	182.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMGCL	152.8	100%	99%	HMG-CoA lyase deficiency, 246450
HNMT	169.7	100%	99%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPH2	180.6	100%	100%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	78.5	87%	81%	Au-Kline syndrome, 616580
HNRNPU	153.2	99%	98%	Epileptic encephalopathy, early infantile, 54, 617391
HOXA1	193.3	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536

HPD	159.2	100%	99%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	82.8	96%	89%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HRAS	204.3	100%	99%	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	119.7	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSPA9	105.5	93%	87%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	105.4	98%	93%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HUWE1	107.1	99%	96%	Mental retardation, X-linked syndromic, Turner type, 300706
IARS	166.1	99%	99%	Growth retardation, intellectual developmental disorder, hypotonia and hepatopathy, 617093
IDS	113.9	99%	97%	Mucopolysaccharidosis II, 309900
IDUA	120.5	92%	86%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	72.1	92%	80%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	143.7	99%	98%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFT172	125.7	100%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT81	102.1	90%	85%	No OMIM phenotype Asphyxiating thoracic dystrophy (Duran (2016) Sci Rep 6, 34232) Short-rib polydactyly syndrome (Duran (2016) Sci Rep 6, 34232) Ciliopathy (Perrault (2015) J Med Genet 52,657)
IGBP1	126.1	99%	96%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	157.6	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	57.4	83%	72%	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	135.1	100%	98%	Mental retardation, X-linked 21/34, 300143
IMPA1	86.8	98%	91%	Mental retardation, autosomal recessive 59, 617323
INPP5E	109	97%	92%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	119	99%	98%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
IQSEC2	72.6	94%	86%	Mental retardation, X-linked 1/78, 309530
ISPD	130.8	97%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGA7	145.6	99%	96%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPR1	177.7	100%	99%	Gillespie syndrome,206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	123.5	100%	99%	Isovaleric acidemia, 243500
JAG1	160.7	99%	98%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAM3	158.3	99%	98%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	174.3	100%	99%	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	169.3	99%	99%	{Coronary heart disease, susceptibility to, 5}, 608901
KANK1	163.5	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	90.1	95%	89%	Koolen-De Vries syndrome, 610443
KAT6A	202	100%	99%	Mental retardation, autosomal dominant 32, 616268
KAT6B	194.1	99%	99%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	166.2	100%	100%	Lissencephaly 6, with microcephaly, 616212
KCNA2	178.9	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366

KCNA4	167.7	100%	100%	No OMIM phenotype Abnormal striatum, congenital cataract and intellectual disability (Kaya (2016) J Med Genet 53,786)
KCNB1	150.8	100%	99%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC3	148.5	72%	58%	Spinocerebellar ataxia 13, 605259
KCNH1	195.4	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	219.9	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	281.8	100%	100%	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ6	192.3	100%	100%	Keppen-Lubinsky syndrome, 614098
KCNK9	214.4	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	114.3	98%	95%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	118.7	99%	96%	Seizures, benign neonatal, type 2, 121201
KCNQ5	174.3	98%	96%	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
KCNT1	133.4	95%	94%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	157.8	94%	93%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	166	99%	97%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5C	123.7	98%	95%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	118	95%	89%	Kabuki syndrome 2, 300867
KIAA0226	117.9	98%	97%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA0586	133.4	98%	94%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1033	114.7	97%	92%	?Mental retardation, autosomal recessive 43, 615817
KIAA1109	175.4	99%	98%	no OMIM phenotype Dandy-Walker malformation,hydrocephalus,flexed deformity,club feet,micrognathia and pleural effusion (Alazami (2015) Cell Rep 10,148)

				?Schizophrenia (Gulsuner (2013) Cell 154,518)
KIAA1279	191.4	100%	100%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	176.1	100%	99%	Mental retardation, X-linked 98, 300912
KIDINS220	193.1	100%	99%	Spastic paraplegia, intellectual disability, nystagmus and obesity, 617296
KIF11	98	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	130.8	99%	97%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF2A	126.5	99%	95%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	101.2	95%	90%	?Mental retardation, X-linked 100, 300923
KIF5C	133.7	99%	98%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	95.3	95%	89%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KIRREL3	157.3	99%	99%	Mental retardation, autosomal dominant 4, 612581
KLHL15	190.8	100%	99%	Mental retardation,X-linked 103,300982
KMT2A	165.3	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2B	139.5	94%	92%	Dystonia 28, childhood-onset, 617284
KMT2C	191.7	91%	89%	No OMIM phenotype Kleefstra syndrome (Kleefstra (2012) Am J Hum Genet 91,73) ?Colorectal cancer and acute myeloid leukaemia (Li (2013) Blood 121, 1478) ?Nasopharyngeal carcinoma (Sasaki (2015) Cancer Epidemiol Biomarkers prev)
KMT2D	158.6	100%	99%	Kabuki syndrome 1, 147920
KPTN	122.9	100%	99%	Mental retardation, autosomal recessive 41, 615637
KRAS	89.6	99%	99%	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	152.5	99%	97%	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	139.7	98%	97%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	154	100%	99%	Poretti-Boltshauser syndrome, 615960
LAMA2	176.7	100%	99%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMC3	151.5	98%	96%	Cortical malformations, occipital, 614115
LAMP2	120.4	93%	91%	Danon disease, 300257
LARGE	142.4	100%	99%	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	68.5	82%	70%	Alazami syndrome, 615071
LAS1L	107	99%	97%	Wilson-Turner syndrome, 309585
LIAS	175	100%	99%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	207.5	100%	99%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LINS	158	99%	99%	Mental retardation, autosomal recessive 27, 614340
LMAN2L	144.1	100%	98%	?Mental retardation, autosomal recessive, 52,616887
LONP1	164.9	98%	97%	CODAS syndrome, 600373
LRP2	205.6	100%	99%	Donnai-Barrow syndrome, 222448
LRPPRC	157.1	99%	97%	Leigh syndrome, French-Canadian type, 220111
LZTFL1	142	99%	98%	Bardet-Biedl syndrome 17, 615994
MAF	67	79%	74%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAGEL2	136.6	100%	100%	Schaaf-Yang syndrome, 615547
MAGT1	118.7	98%	97%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAN1B1	162.6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	134.6	98%	95%	Mannosidosis, alpha-, types I and II, 248500
MANBA	153.6	99%	98%	Mannosidosis, beta, 248510
MAOA	130.2	100%	99%	Brunner syndrome, 300615

				{Antisocial behavior},300615
MAP2K1	106.2	99%	97%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	122.2	99%	95%	Cardiofaciocutaneous syndrome 4, 615280
MAPRE2	221.3	100%	99%	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	160.1	99%	99%	3MC syndrome 1, 257920
MAT1A	206.1	99%	97%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	204.8	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBOAT7	102.1	99%	97%	Mental retardation, autosomal recessive 57, 617188
MBTPS2	147.5	99%	98%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MCCC1	176.8	100%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	159.5	100%	99%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	168.5	99%	97%	Mucolipidosis IV, 252650
MCPH1	156.6	99%	98%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	121.1	98%	97%	Epileptic encephalopathy, early infantile, 51, 617339
MECP2	99.4	99%	95%	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	120.5	99%	96%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	111.4	98%	94%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	141.7	99%	99%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	156.8	99%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	162.9	99%	97%	Mental retardation, autosomal recessive 18, 614249
MED25	124.1	98%	95%	Basel-Vanagait-Smirin-Yosef syndrome, 616449

				?Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	150.6	99%	96%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEIS2	154	100%	100%	No OMIM phenotype Heart defect, cleft palate & intellectual disability (Louw (2015) Am J Med Genet A 167, 1142) Orofacial clefting & delayed motor development (Johansson (2014) Am J Med Genet A 164, 1622)
METTL23	154	100%	100%	Mental retardation, autosomal recessive 44, 615942
MFSD2A	134.8	99%	99%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	143.4	100%	99%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	177.1	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MICU1	134.3	97%	93%	Myopathy with extrapyramidal signs, 615673
MID1	177.2	99%	98%	Opitz GBBB syndrome, type I, 300000
MID2	151.2	99%	98%	?Mental retardation, X-linked 101, 300928
MKKS	216.9	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.6	99%	99%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MLC1	114.2	100%	99%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	94.5	95%	91%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	222.3	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMACHC	227.3	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94.4	90%	80%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	92.7	98%	92%	Molybdenum cofactor deficiency A, 252150
MOCS2	183	99%	99%	Molybdenum cofactor deficiency B, 252160
MOGS	128	99%	98%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	129.4	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPDZ	171.5	98%	97%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPLKIP	103.8	99%	91%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRPL3	74.4	92%	82%	Combined oxidative phosphorylation deficiency 9, 614582
MRPS22	167.4	97%	93%	Combined oxidative phosphorylation deficiency 5, 611719
MTFMT	148.9	99%	96%	Combined oxidative phosphorylation deficiency 15, 614947

MTHFR	161.7	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTOR	152.6	100%	99%	Smith-Kingsmore syndrome, 616638
MTR	174.9	99%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	152.5	99%	99%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	154.5	99%	98%	Methylmalonic aciduria, mut(0) type, 251000
MVK	167.2	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYCN	117	95%	86%	Feingold syndrome, 164280
MYH9	152.2	99%	98%	Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249
MYO5A	138.4	99%	98%	Griscelli syndrome, type 1, 214450
MYT1L	199.4	100%	99%	Mental retardation, autosomal dominant 39, 616521
NAA10	109.4	99%	96%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NACC1	193.4	100%	99%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties and delayed brain myelination, 617393
NAGA	159.7	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	131.8	94%	91%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NALCN	147	99%	98%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	115	100%	99%	Sponyloepimetaphyseal dysplasia, Genevieve type,610442

NARS2	153	97%	97%	Combined oxidative phosphorylation deficiency 24, 616239
NBN	106	99%	98%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NDE1	106.3	100%	99%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDP	126.5	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	231.1	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	229	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	95.2	99%	95%	Mitochondrial complex I deficiency, 252010
NDUFA12	175	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	146.8	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF3	130.4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	108.8	99%	98%	Mitochondrial complex 1 deficiency, 252010
NDUFS1	165.9	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS2	121.6	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	149.4	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	200.6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	146.2	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS7	141.6	100%	99%	Leigh syndrome, 256000
NDUFS8	146.8	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	158	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	88.7	90%	73%	Mitochondrial complex I deficiency, 252010
NECAP1	135.2	100%	100%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	159.4	99%	99%	Periventricular nodular heterotopia 7, 617201
NEU1	20.4	72%	43%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NF1	146.2	93%	91%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321

				Watson syndrome, 193520
NFATC1	137.1	99%	97%	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	167.2	100%	99%	No OMIM phenotype Brain malformation and urinary tract defect (Negishi (2015) Hum Genome Var 2) Bipolar disorder & depression (Mikhail (2011) Am J Med Genet A 155,2386) Central nervous system malformations (Koehler (2010) Eur J Pediatr 169,463) Intellectual disability with macrocephaly (Labonne (2016) Mol Cytogenet 9,24)
NFIX	166.6	98%	96%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NGLY1	162.3	100%	99%	Congenital disorder of deglycosylation, 615273
NHS	138.9	96%	93%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIPBL	142.5	97%	95%	Cornelia de Lange syndrome 1, 122470
NKX2-1	58.2	99%	92%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550
NLGN3	156.8	100%	99%	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	189	99%	98%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRP3	162.9	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NONO	110.4	99%	96%	Mental retardation, X-linked, syndromic 34, 300967
NPC1	153.4	99%	99%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	148.6	100%	99%	Niemann-pick disease, type C2, 607625
NPHP1	154.6	99%	98%	Joubert syndrome 4, 609583

				Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NR2F1	216.5	99%	98%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRAS	203.3	100%	100%	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
NRXN1	193.5	99%	98%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	181.1	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	196.4	100%	99%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	133.7	95%	94%	Mental retardation, autosomal recessive 5, 611091
NTRK1	160.6	99%	97%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUP62	113.7	100%	99%	Striatonigral degeneration, infantile, 271930
OAT	96.2	80%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	239.3	100%	100%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	140.5	99%	98%	Dent disease 2, 300555 Lowe syndrome, 309000
ODC1	156.9	100%	99%	{Colonic adenoma recurrence, reduced risk of}, 114500
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	103.5	99%	97%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	129.5	99%	98%	Meier-Gorlin syndrome 1, 224690
OTC	128.2	99%	99%	Ornithine transcarbamylase deficiency, 311250
PACS1	130.4	97%	95%	Schuss-Hoeijmakers-syndrome, 615009

PAFAH1B1	125	92%	87%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	186.6	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	94.1	98%	93%	Mental retardation, X-linked 30/47, 300558
PANK2	178.4	99%	98%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	174.4	100%	100%	No OMIM phenotype Intellectual disability, sensorineural hearing loss, skeletal defects and primary ovarian failure (Shao (2016) J Biol Chem 291,12432)
PAX1	138	90%	84%	?Otofaciocervical syndrome 2, 615560
PAX6	135.8	100%	100%	Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229 ?Morning glory disc anomaly, 120430
PAX8	101.3	99%	98%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PC	166.1	99%	97%	Pyruvate carboxylase deficiency, 266150
PCCA	120.1	98%	93%	Propionicacidemia, 606054
PCCB	167.8	98%	96%	Propionicacidemia, 606054
PCDH19	219.8	99%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	121.5	99%	97%	no OMIM phenotype ?Developmental disorder (Fitzgerald (2015) Nature 519,223)
PCLO	191.9	99%	99%	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	128.9	99%	96%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	132.3	98%	96%	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDHA1	112.7	98%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDP1	197.1	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	140.2	93%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	146.3	98%	95%	Coenzyme Q10 deficiency, primary, 3, 614652

PEPD	127	99%	98%	Prolidase deficiency, 170100
PET100	98.1	98%	86%	Mitochondrial complex IV deficiency, 220110
PEX1	139.9	98%	97%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	124.7	97%	92%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	103.8	100%	99%	Peroxisome biogenesis disorder 14B, 614920
PEX12	157.9	100%	99%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	240	100%	99%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	146.3	97%	93%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	118.8	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	187.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	87.2	100%	99%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	129.6	99%	97%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	132.1	99%	98%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	95.9	91%	85%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	152.7	90%	87%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGAP1	125.3	97%	91%	Mental retardation, autosomal recessive 42, 615802
PGAP2	173.9	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	126.1	98%	94%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	57.8	92%	80%	Phosphoglycerate kinase 1 deficiency, 300653
PHF6	74.9	94%	86%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	102.5	99%	97%	Mental retardation syndrome, X-linked, Siderius type, 300263

PHGDH	138	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	156.2	97%	94%	No OMIM phenotype Glaucoma,primary congenital (Lee (2011) Mol Vis 17,3583) Intellectual disability (de Ligt (2012) N Engl J Med 367,1921)
PI4KA	118	93%	89%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIGA	97	94%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	117.8	99%	94%	No OMIM phenotype ?Hydrops fetalis, nonimmune (Shamseldin (2015) Genome Biol 16,116)
PIGG	192.9	100%	99%	Mental retardation,autosomal recessive 53,616917
PIGL	125.2	100%	99%	CHIME syndrome, 280000
PIGN	130.2	99%	94%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	143.9	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	195.8	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	168	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	186	100%	99%	?Hyperphosphatasia with mental retardation syndrome 5, 616025
PIGY	121.8	100%	99%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	155.4	100%	99%	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	104.3	90%	87%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	135.5	99%	97%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	173.5	100%	99%	Epileptic encephalopathy, early infantile, 12, 613722

PLP1	155	99%	98%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLXND1	135.4	97%	94%	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	171.3	100%	99%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	135.9	98%	95%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKP	100.7	99%	97%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	147.3	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POC1A	144.7	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POGZ	172.9	99%	98%	White-Sutton syndrome, 616364
POLG	128.1	100%	99%	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	153	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	163.8	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	136.7	99%	97%	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
POMGNT2	258.7	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	223.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094
POMT1	184.3	99%	98%	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	117.5	99%	96%	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	142.4	100%	99%	Focal dermal hypoplasia, 305600

POU1F1	141.9	99%	97%	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	36.4	73%	61%	No OMIM phenotype ?Intellectual disability (Dheedene (2014) Mol Syndromol 5,32)
PPOX	104.3	99%	97%	Porphyria variegata, 176200
PPP1CB	126.2	100%	99%	No OMIM phenotype Intellectual disability, syndromic (Ma (2016) Hum Genet 135, 1399)
PPP1R15B	156	99%	97%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP2R1A	158.2	93%	93%	Mental retardation, autosomal dominant 36, 616362
PPP2R5B	124.1	100%	98%	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24, 4775)
PPP2R5C	122.9	96%	91%	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24,4775)
PPP2R5D	166.9	100%	99%	Mental retardation, autosomal dominant 35, 616355
PPT1	203.5	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	180.7	97%	96%	Renpenning syndrome, 309500
PRKAR1A	104.2	97%	92%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRMT7	154.8	100%	99%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	96.4	89%	83%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	178.9	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
PRSS12	178.3	99%	99%	Mental retardation, autosomal recessive 1, 249500
PSAP	129.6	99%	99%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSEN1	156.6	99%	97%	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
PTCH1	127.7	98%	96%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCHD1	169.5	100%	99%	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	162.9	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	169.6	100%	99%	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTRH2	305.6	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	161.8	100%	99%	No OMIM phenotype ?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry)
PTS	133.3	99%	96%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	197.3	99%	98%	Verheij syndrome, 615583
PURA	135.6	98%	93%	Mental retardation, autosomal dominant 31, 616158
PUS1	123.1	99%	97%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	232.8	100%	100%	?Mental retardation, autosomal recessive 55, 617051
PUS7	182	99%	98%	No OMIM phenotype

PVRL1	166.5	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PYCR1	103.5	99%	95%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	142.2	99%	96%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	163.6	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	91.4	100%	99%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	108.6	99%	91%	Warburg micro syndrome 3, 614222
RAB27A	171.6	100%	100%	Griscelli syndrome, type 2, 607624
RAB39B	125.7	100%	99%	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RAB3GAP1	155.9	99%	99%	Warburg micro syndrome 1, 600118
RAB3GAP2	116.8	99%	96%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	167.2	100%	99%	No OMIM phenotype ?Martin-Probst syndrome (Bedoyan (2012) J Med Genet 49, 332)
RAC1	141.2	99%	94%	No OMIM phenotype ?Radial ray defect (Vergult (2013) Genet Med 15, 195) {Ulcerative colitis, increased risk, association with} (Muise (2011) Gastroenterology 141,633)
RAD21	109	99%	97%	Cornelia de Lange syndrome 4, 614701
RAF1	144.9	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAI1	159.2	100%	99%	Smith-Magenis syndrome, 182290
RARB	147.3	100%	100%	Microphthalmia, syndromic 12, 615524
RARS2	137.6	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RBFOX1	182.7	99%	98%	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	130.8	99%	98%	TARP syndrome, 311900
RBM28	180.5	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079

RBPJ	94.2	95%	88%	Adams-Oliver syndrome 3, 614814
RCBTB1	135.4	100%	99%	[Beta-glycopyranoside tasting] {Alcohol dependence, susceptibility to}, 103780
RELN	184.8	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
RERE	86	96%	93%	Neurodevelopmental disorder with or without anomalies of the brain,eye or heart, 616975
REV3L	172.2	99%	98%	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	114.8	99%	98%	Congenital disorder of glycosylation, type In, 612015
RHEB	40.3	87%	73%	No OMIM phenotype
RIT1	190.3	100%	100%	Noonan syndrome 8, 615355
RLIM	142.8	100%	98%	Mental Retardation, X-linked 61, 300978
RMND1	168.4	99%	98%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	157.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	128.3	98%	92%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	244.3	99%	98%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	99.3	93%	86%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	156.8	100%	100%	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	181.7	100%	99%	Tenorio syndrome, 616260
ROGDI	127.1	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPGRIP1L	160.8	96%	95%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	94.2	98%	91%	{Autism, susceptibility to, X-linked 5}, 300847
RPS6KA3	97.9	96%	89%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RSPRY1	186.1	100%	100%	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723
RTEL1	137.2	99%	97%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190

				Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTTN	144.1	98%	96%	Microcephaly,short stature,and polymicrogyria with seizures,614833
RUSC2	191.4	100%	99%	No OMIM phenotype Intellectual disability and secondary microcephaly (Alwadei (2016) Dev Med Child Neurol epub, epub)
SALL1	144	99%	98%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SATB2	122.4	99%	94%	Glass syndrome, 612313
SBDS	231.2	100%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SC5D	237.6	99%	99%	Lathosterolosis, 607330
SCN1A	170.2	99%	99%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	189.8	97%	96%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	186.5	99%	98%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN8A	209.5	99%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SCO1	122.6	98%	94%	Mitochondrial complex IV deficiency, 220110
SCO2	126.5	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	123.2	84%	79%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SEMA3E	179.1	100%	99%	?CHARGE syndrome, 214800
SEPSECS	198.4	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	139.3	99%	96%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	142.5	97%	96%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	137.2	98%	96%	No OMIM phenotype

				Schizophrenia (Takata (2014) Neuron 82, 723)
SETD2	182.8	100%	99%	Luscan-Lumish syndrome, 616831
SETD5	200.5	100%	99%	Mental retardation, autosomal dominant 23, 615761
SF1	91.3	87%	81%	No OMIM phenotype
SGSH	140.9	96%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SHANK2	139.8	99%	99%	{Autism susceptibility 17}, 613436
SHANK3	90.5	86%	76%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	114.4	99%	95%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	150.5	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	113.4	100%	98%	?Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	111.9	98%	96%	Epileptic encephalopathy, early infantile, 30, 616341
SIL1	175.7	99%	97%	Marinesco-Sjogren syndrome, 248800
SIN3A	158.1	99%	98%	Witteveen-Kolk syndrome, 613406
SIX3	172.7	99%	98%	Holoprosencephaly-2, 157170 Schizencephaly, 269160
SKI	96.2	98%	96%	Shprintzen-Goldberg syndrome, 182212
SLC12A6	162.9	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	167.9	100%	99%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	70.9	96%	87%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	142.3	99%	95%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	185.8	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	215.5	100%	99%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A2	136.1	99%	99%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	190.5	99%	98%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A12	183.5	99%	99%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	238.4	98%	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	120.5	99%	97%	Epileptic encephalopathy, early infantile, 3, 609304

SLC2A1	191.5	100%	99%	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	152.6	98%	94%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	169.7	100%	99%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	111.5	99%	97%	Congenital disorder of glycosylation, type IIm, 300896
SLC35C1	244.4	99%	98%	Congenital disorder of glycosylation, type IIc, 266265
SLC39A12	128.2	99%	97%	No OMIM phenotype
SLC39A8	160.7	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC4A4	159.1	99%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A1	167	100%	99%	Myoclonic-atonic epilepsy, 616421
SLC6A17	219.8	100%	99%	Mental retardation, autosomal recessive 48, 616269
SLC6A3	150.7	100%	99%	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	59	91%	81%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	114.5	100%	99%	Lysinuric protein intolerance, 222700
SLC9A6	119.2	98%	94%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	136.5	99%	99%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMARCA2	127.7	97%	96%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	165.4	100%	99%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	265.7	100%	100%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCC2	123.3	99%	96%	No OMIM phenotype ?Ivemark syndrome (Carss (2014) Hum Mol Genet 23,3269) ?Autism (Neale (2012) Nature 485,242)
SMARCE1	97.3	97%	90%	Coffin-Siris syndrome 5,616938

				{Meningioma, familial, susceptibility to}, 607174
SMC1A	114.2	100%	98%	Cornelia de Lange syndrome 2,300590
SMC3	96.1	94%	88%	Cornelia de Lange syndrome 3, 610759
SMOC1	130.7	99%	97%	Microphthalmia with limb anomalies, 206920
SMPD1	151.8	99%	98%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	79.4	89%	77%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	144.9	100%	100%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	149.1	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	164.9	99%	97%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRPN	128.6	100%	99%	Prader-Willi syndrome, 176270
SNX14	89.1	96%	88%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	136.1	95%	89%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	158.6	98%	95%	ZITK syndrome, 617140
SOS1	120.1	98%	95%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	115.8	99%	97%	Noonan syndrome 9, 616559
SOX10	74.6	98%	95%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	129.8	99%	98%	Mental retardation, autosomal dominant, 27, 615866
SOX2	127.1	99%	98%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	47	94%	81%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	125.3	100%	99%	Lamb-Shaffer syndrome, 616803
SPAST	82.7	97%	88%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	153.8	100%	99%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG11	145	99%	98%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPOCK1	126.9	100%	99%	No OMIM phenotype Developmental delay and microcephaly (Dhamija (2014) Eur J Med Genet 57,181)
SPRED1	196.7	99%	96%	Legius syndrome, 611431

SPTAN1	137.5	99%	99%	Epileptic encephalopathy, early infantile, 5, 613477
SPTBN2	127.6	99%	99%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SRCAP	165.3	99%	98%	Floating-Harbor syndrome, 136140
SRD5A3	172	100%	99%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	81.4	99%	97%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SSR4	106.7	100%	99%	?Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	195.5	100%	99%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	144.8	95%	94%	Amish infantile epilepsy syndrome, 609056
STAG1	134.7	99%	97%	No OMIM phenotype ?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub) ?Schizophrenia (Li (2016) Mol Psychiatry 21,290)
STAMBP	125	99%	97%	Microcephaly-capillary malformation syndrome, 614261
STIL	205.1	99%	99%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	120.5	100%	99%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STT3A	181.8	100%	100%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	156.7	99%	98%	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	191.4	100%	99%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	146.2	100%	99%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	78.2	94%	86%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	120	100%	99%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUMF1	137.1	99%	94%	Multiple sulfatase deficiency, 272200
SUOX	221.3	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	98.6	89%	88%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYN1	73.7	84%	71%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	71.1	95%	86%	No OMIM phenotype ?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)
SYNE1	172	99%	99%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998

				Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNGAP1	77.1	95%	86%	Mental retardation, autosomal dominant 5, 612621
SYP	81.3	99%	96%	Mental retardation, X-linked 96, 300802
SYT14	208	96%	87%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	158.8	99%	99%	Epileptic encephalopathy, early infantile, 18, 615476
TAF1	133.9	99%	97%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF2	139.3	99%	97%	Mental retardation, autosomal recessive 40, 615599
TAT	140.9	100%	100%	Tyrosinemia, type II, 276600
TBC1D20	165.4	94%	94%	Warburg micro syndrome 4, 615663
TBC1D24	203.2	100%	99%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOOR syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	118.4	99%	97%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	173.7	98%	95%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	162.8	99%	99%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	111.7	97%	93%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3,616900
TBL1XR1	103.7	95%	82%	Mental retardation, autosomal dominant 41,616944 Piermont syndrome,602342
TBP	137.1	100%	99%	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBR1	133.6	99%	98%	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	164.6	100%	100%	No OMIM phenotype Autism spectrum disorder (Babbs (2014) J Med Genet 51,737)
TCF4	150.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	168.1	99%	97%	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCN2	201.5	100%	100%	Transcobalamin II deficiency, 275350

TCTN3	133.6	100%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDP2	203.7	100%	99%	Spinocerebellar ataxia, autosomal recessive, 616949
TECPR2	169.7	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	108.3	99%	98%	Mental retardation, autosomal recessive 14, 614020
TELO2	121.1	98%	95%	You-Hoover-Fong syndrome, 616954
TFAP2A	126.8	100%	99%	Branchiooculofacial syndrome, 113620
TGFBR1	204.7	94%	93%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	212.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	151.7	100%	99%	Holoprosencephaly-4, 142946
TH	87.6	97%	93%	Segawa syndrome, recessive, 605407
THOC2	95	97%	91%	Mental retardation, X-linked 12/35, 300957
THOC6	269.9	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	204.9	99%	99%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	43.7	88%	71%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TINF2	208.7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLK2	111.9	98%	94%	No OMIM phenotype ?Schizophrenia (Gulsuner (2013) Cell 154,518) ?Autism spectrum disorder (Li (2016) Mol Psychiatry 21,290)
TMCO1	114.3	100%	99%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM165	136.2	99%	98%	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	105.5	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	126	99%	98%	Joubert syndrome 14, 614424
TMEM240	136.1	100%	99%	Spinocerebellar ataxia 21, 607454
TMEM67	93.3	95%	89%	COACH syndrome, 216360 Joubert syndrome 6, 610688

				Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	172	96%	93%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	110.9	99%	96%	{Autism,susceptibility to,X-linked 6}, 300872
TNIK	139.9	100%	99%	Mental retardation, autosomal recessive 54, 617028
TOE1	157.9	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
TPI1	104.9	98%	95%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	165	99%	97%	Thyroid dyshormonogenesis 2A, 274500
TPP1	155.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAPPC11	156	99%	97%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC9	164.3	100%	99%	Mental retardation, autosomal recessive 13, 613192
TREX1	302.9	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	147.3	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIO	158.3	98%	96%	Mental retardation, autosomal dominant 44,617061
TRIP12	164.3	99%	99%	No OMIM phenotype ?Autism (Iossifov (2012) Neuron 74,285)
TRMT1	120.9	99%	98%	No OMIM phenotype Intellectual disability (Davarniya (2015) PLoS One 10,e0129631)
TRMT10A	176.3	100%	99%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TSC1	140.4	99%	97%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	150.1	99%	99%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	97.3	99%	93%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN54	98.8	96%	93%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSHB	372.8	100%	100%	Hypothryoidism, congenital, nongoitrous 4, 275100

TSPAN7	137.2	100%	99%	Mental retardation, X-linked 58, 300210
TTC19	105.8	90%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	159.2	99%	98%	Trichohepatoenteric syndrome 1, 222470
TTC8	118.3	99%	98%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	109.9	100%	99%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	115.3	99%	97%	Lissencephaly 3, 611603
TUBA8	171.3	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB	21.1	77%	49%	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB2B	104.4	100%	100%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	212.4	99%	98%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	135.6	96%	95%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	190.6	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	149	99%	96%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	176.6	99%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	166.8	100%	99%	Mental retardation, autosomal recessive 7, 611093
TWIST1	135.7	94%	85%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Saethre-Chotzen syndrome, 101400
UBA5	103.4	97%	86%	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia, autosomal recessive 24, 617133
UBE2A	110.5	99%	98%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	117.2	99%	96%	Angelman syndrome, 105830
UBE3B	145.7	100%	99%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	144.2	99%	98%	Johanson-Blizzard syndrome, 243800
UNC80	151.4	99%	99%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	184.6	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	68.3	95%	87%	Mental retardation, X-linked, syndromic 14, 300676
UQCRQ	180.8	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	164	100%	99%	?Urocanase deficiency, 276880

USP27X	258.4	100%	100%	No OMIM phenotype Intellectual disability (Hu (2015) Mol Psychiatry epub,epub)
USP7	116	94%	90%	No OMIM phenotype ?Autism spectrum disorder (Levy (2011) Neuron 70,886)
USP9X	135.2	98%	94%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
VLDLR	217.6	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	170.6	100%	99%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	170.3	99%	97%	Cohen syndrome, 216550
VPS37A	86.4	93%	76%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	145	91%	90%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	157.2	99%	97%	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	163	100%	99%	?Spinocerebellar ataxia,autosomal recessive 22,616948
WAC	178.2	99%	98%	Desanto-Shinawi syndrome, 616708
WDR13	140.4	99%	99%	No OMIM phenotype Intellectual disability,X-linked (Whibley (2010) Am J Hum Genet 87,173)
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR45	85.2	95%	89%	Neurodegeneration with brain iron acculation 5, 300894
WDR62	178	99%	99%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	147.2	100%	99%	Galloway-Mowat syndrome, 251300
WDR81	187.7	100%	99%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	260.1	99%	99%	Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296 ?Cataract 41, 116400 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WWOX	143.3	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XPA	69.2	97%	89%	Xeroderma pigmentosum, group A, 278700
XPNPEP3	142.9	98%	97%	Nephronophthisis-like nephropathy 1, 613159

XYLT1	144.5	92%	88%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	112.2	90%	84%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	125	98%	95%	?Optic atrophy 11, 617302
YWHAE	133.3	99%	97%	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	146.1	100%	99%	No OMIM phenotype Mental retardation (Vissers (2010) Nat Genet 42,1109)
ZBTB16	165.9	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	227.2	99%	99%	?Mental retardation, autosomal dominant 22, 612337
ZBTB20	226.5	100%	100%	Primrose syndrome, 259050
ZBTB24	213.6	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC3H14	205.6	99%	98%	Mental retardation, autosomal recessive 56, 617125
ZC4H2	93.9	99%	97%	Wieacker-Wolff syndrome, 314580
ZDHHC15	100.8	99%	96%	?Mental retardation, X-linked 91, 300577
ZDHHC9	63.6	99%	90%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	195.1	100%	99%	Mowat-Wilson syndrome, 235730
ZFYVE26	130.7	100%	99%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	213	100%	100%	Craniosynostosis 6, 616602
ZIC2	116.8	92%	84%	Holoprosencephaly-5, 609637
ZMYND11	146.6	100%	99%	Mental retardation, autosomal dominant 30, 616083
ZNF292	164.6	99%	98%	No OMIM phenotype ?Autism (Neale (2012) Nature 485,242)
ZNF407	179.2	99%	98%	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	127.2	100%	99%	Mental retardation, X-linked 89, 300848
ZNF592	139.4	100%	99%	Spinocerebellar ataxia, autosomal recessive 5, 251300

ZNF674	140.4	100%	100%	Mental retardation, X-linked 92, 300851
ZNF711	155.1	99%	97%	Mental retardation, X-linked 97, 300803
ZNF81	93.1	99%	96%	Mental retardation, X-linked 45, 300498
ZSWIM6	176.5	94%	91%	Acromelic frontonasal dysostosis, 603671

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 : April 14th 2017
This list is accurate for panel version DG 2.9 and DG 2.10

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