INTELLECTUAL DISABILITY GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
A2ML1	154.8	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	144.7	99%	99%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	103	100%	99%	GABA-transaminase deficiency, 613163
ABCC9	177.1	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	96	77%	68%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	155.2	99%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	265.7	99%	99%	Chanarin-Dorfman syndrome, 275630
ACAD9	154	99%	96%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACO2	138.6	96%	92%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	186.3	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	146.5	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	127.6	96%	91%	Mental retardation, X-linked 63, 300387
ACTB	134.1	98%	93%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	139.4	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACVR1	180.1	100%	99%	Fibrodysplasia ossificans progressiva, 135100
ACY1	155.9	99%	98%	Aminoacylase 1 deficiency, 609924
ADAR	131.7	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	83.4	98%	95%	Mental retardation, autosomal recessive 36, 615286

ADCK3	146	99%	98%	Coenzyme Q10 deficiency, primary, 4, 612016
ADK	106.1	98%	93%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	280.5	100%	99%	Helsmoortel-van der Aa syndrome, 615873
ADSL	205.9	100%	99%	Adenylosuccinase deficiency, 103050
AFF2	156.1	99%	99%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	130	99%	96%	CHOPS syndrome, 616368
AGA	154.1	100%	100%	Aspartylglucosaminuria, 208400
AGPAT2	120.1	98%	92%	Lipodystrophy, congenital generalized, type 1, 608594
AHCY	138.6	100%	99%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	136	99%	97%	Xia-Gibbs syndrome, 615829
AHI1	151.9	98%	94%	Joubert syndrome-3, 608629
AIFM1	133.5	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	96.4	97%	90%	Leukodystrophy, hypomyelinating, 3, 260600
AK1	130.7	99%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AKT3	83.7	97%	89%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	143	100%	99%	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	157.4	100%	99%	Sjogren-Larsson syndrome, 270200
ALDH4A1	130.3	99%	98%	Hyperprolinemia, type II, 239510
ALDH5A1	92.3	90%	82%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	60.3	53%	49%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	182.7	100%	99%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	167.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	107.4	98%	94%	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	119.8	100%	99%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	125.8	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	104.1	95%	92%	Congenital disorder of glycosylation, type Ic, 603147
ALG9	136.2	99%	98%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210

ALMS1	197.7	99%	99%	Alstrom syndrome, 203800
ALX1	171.3	99%	96%	?Frontonasal dysplasia 3, 613456
ALX4	132.6	96%	89%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMPD2	145.3	99%	98%	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	174	100%	99%	Glycine encephalopathy, 605899
ANK3	187.9	99%	99%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	121.4	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	107.9	97%	93%	KBG syndrome, 148050
ANO10	126.6	99%	96%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	141.9	98%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S2	75	79%	68%	Mental retardation, X-linked syndromic 5, 304340
AP3B1	111.5	97%	91%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	166	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	117.3	99%	96%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	125.9	99%	97%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78.3	74%	68%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	136.6	93%	90%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	179.2	99%	98%	Periventricular heterotopia with microcephaly, 608097
ARG1	172.1	100%	100%	Argininemia, 207800
ARHGAP31	128.3	99%	98%	Adams-Oliver syndrome 1, 100300
ARHGEF6	168.8	95%	93%	Mental retardation, X-linked 46, 300436
ARHGEF9	109.7	99%	98%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	155.1	95%	90%	Coffin-Siris syndrome 2, 614607
ARID1B	157	95%	90%	Coffin-Siris syndrome 1, 135900
ARL13B	100.4	99%	92%	Joubert syndrome 8, 612291
ARL6	99	97%	91%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900

ARSE	108.8	98%	93%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	39.1	82%	70%	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
ASL	118.4	99%	98%	Argininosuccinic aciduria, 207900
ASNS	97.8	97%	91%	Asparagine synthetase deficiency, 615574
ASPA	151.7	99%	92%	Canavan disease, 271900
ASPM	110.9	97%	92%	Microcephaly 5, primary, autosomal recessive, 608716
ASXL1	168.4	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	178.5	99%	99%	Bainbridge-Ropers syndrome, 615485
ATCAY	166.6	100%	99%	Ataxia, cerebellar, Cayman type, 601238
ATIC	134.7	99%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A2	209.6	100%	100%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP2A2	197.2	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP6AP2	55.4	83%	63%	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	159.8	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	157.3	99%	97%	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	114.3	100%	99%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	160.3	98%	96%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	94.5	97%	93%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040

				Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	89.3	98%	92%	3-methylglutaconic aciduria, type I, 250950
AUTS2	115.2	96%	95%	Mental retardation, autosomal dominant 26,615834
B3GALNT2	134.9	92%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALTL	113.5	96%	92%	Peters-plus syndrome, 261540
B3GNT1	126	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B4GALT1	132.4	99%	99%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	116.3	96%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BBS1	162.3	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	179.1	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	225	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	210.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	163.9	99%	97%	Bardet-Biedl syndrome 4, 615982
BBS5	122.1	95%	89%	Bardet-Biedl syndrome 5, 615983
BBS7	135.5	97%	92%	Bardet-Biedl syndrome 7, 615984
BBS9	124.1	96%	93%	Bardet-Biedl syndrome 9, 615986
BCAP31	74.1	93%	81%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	186.9	99%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	124.4	89%	81%	Maple syrup urine disease, type Ib, 248600
BCL11A	148.5	99%	97%	Intellectual development disorder with persistence of fetal hemoglobin, 617101
BCOR	131.4	99%	97%	Microphthalmia, syndromic 2, 300166
BCS1L	184.4	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	139.6	98%	95%	Bloom syndrome, 210900
BRAF	77	89%	79%	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	112.9	99%	96%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRF1	106.5	97%	93%	Cerebellofaciodental syndrome, 616202
BRWD3	120.6	97%	93%	Mental retardation, X-linked 93, 300659
BSCL2	126.4	100%	99%	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	163.8	100%	99%	Biotinidase deficiency, 253260
BUB1B	164.8	98%	97%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf2	193.6	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
C12orf57	148.2	100%	100%	Temtamy syndrome, 218340
C12orf65	91.3	97%	92%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C2CD3	163.2	95%	95%	?Orofaciodigital syndrome XIV, 615948
C5orf42	136.5	98%	94%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	166.3	98%	93%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	124.8	99%	95%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	120.5	95%	90%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNG2	156.8	100%	100%	Mental retardation, autosomal dominant 10, 614256
CAMTA1	193.2	99%	99%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CASC5	122.3	97%	94%	Microcephaly 4, primary, autosomal recessive, 604321
CASK	116.5	98%	94%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CBL	145.2	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	127.1	97%	92%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	132.5	99%	98%	Mental retardation, autosomal recessive 3, 608443

CC2D2A	137.5	98%	96%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	81.9	98%	92%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC174	126.7	96%	92%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	108.1	97%	91%	Ritscher-Schinzel syndrome 2, 300963
CCDC78	118.4	100%	99%	Myopathy, centronuclear, 4, 614807
CCND2	168.1	100%	99%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CDH15	132.3	99%	95%	Mental retardation, autosomal dominant 3, 612580
CDK5RAP2	146.6	99%	98%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	140.7	98%	96%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1C	35	75%	60%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDON	156.1	99%	99%	Holoprosencephaly 11, 614226
CENPJ	163.1	99%	97%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP135	87.7	96%	87%	?Microcephaly 8, primary, autosomal recessive, 614673
CEP152	195.6	97%	94%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	77.4	88%	77%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	96.7	97%	90%	Joubert syndrome 15, 614464
CHAMP1	184.7	100%	100%	Mental retardation, autosomal dominant 40, 616579
CHD2	148.5	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	118.7	97%	93%	No OMIM phenotype ?Autism (O'Roak (2012) Nature 485,246)
CHD4	150.9	100%	99%	No OMIM phenotype ?Epileptic encephalopathy (Li (2016) Mol Psychiatry 21, 290) {Cancer, increased risk, association with} (Yamada (2015) Genes Chromosomes Cancer 54, 122)
CHD7	161	99%	98%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370

CHD8	172.3	100%	99%	{Autism, susceptibility to, 18}, 615032
CHKB	101.2	99%	96%	Muscular dystrophy, congenital, megaconial type, 602541
CKAP2L	195.2	98%	95%	Filippi syndrome, 272440
CLCN4	133.4	100%	99%	Mental retardation, X-linked 49/15, 300114
CLCNKB	106.8	98%	92%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLIC2	93	98%	95%	?Mental retardation, X-linked, syndromic 32, 300886
CLN3	125.9	98%	94%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	163.1	98%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	142.3	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	252.4	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	152.7	96%	95%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CNNM2	213.2	99%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTNAP2	156.8	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COASY	168	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COG1	136	99%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	139	100%	99%	Congenital disorder of glycosylation, type IIj, 613489
COG5	117.8	97%	93%	Congenital disorder of glycosylation, type IIi, 613612
COG6	92.4	93%	85%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	138.9	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	124.6	99%	96%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	101.8	98%	93%	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	103	98%	94%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519

COL4A3BP	140.6	99%	96%	Mental retardation, autosomal dominant 34, 616351
COLEC11	220.9	100%	100%	3MC syndrome 2, 265050
COQ2	84.5	95%	92%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	94	86%	82%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ9	105.7	99%	98%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	240.2	100%	99%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	105.9	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	174.5	100%	100%	Mitochondrial complex IV deficiency, 220110
CPS1	169.3	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CRADD	126.4	99%	97%	Mental retardation, autosomal recessive 34, 614499
CRBN	157.5	99%	94%	Mental retardation, autosomal recessive 2, 607417
CREBBP	147.6	99%	96%	Rubinstein-Taybi syndrome, 180849
CSNK2A1	138	94%	88%	Okur-Chung neurodevelopmental syndrome, 617062 Glaucoma, primary congenital (Lee (2011) Mol Vis 17,3583)
CSPP1	119.4	99%	96%	Joubert syndrome 21, 615636
CTCF	173.2	99%	97%	Mental retardation, autosomal dominant 21, 615502
CTDP1	112	91%	84%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNB1	189	100%	100%	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTSA	149.8	99%	99%	Galactosialidosis, 256540
CTSD	183.6	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CUBN	141.9	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CUL4B	83.8	96%	90%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CYB5R3	184.9	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
D2HGDH	147.9	97%	95%	D-2-hydroxyglutaric aciduria, 600721

DAG1	221.2	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	137.8	99%	99%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	122.5	97%	92%	Maple syrup urine disease, type II, 248600
DCAF17	110.1	98%	92%	Woodhouse-Sakati syndrome, 241080
DCHS1	167	99%	98%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCPS	159.9	100%	99%	Al-Raqad syndrome, 616459
DCX	137.9	100%	99%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDC	118.6	99%	96%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	184.8	99%	96%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	99.2	80%	73%	Warsaw breakage syndrome, 613398
DDX3X	112.2	98%	95%	Mental retardation, X-linked 102, 300958
DEAF1	149.5	90%	85%	Mental retardation, autosomal dominant 24, 615828
DHCR24	203	100%	99%	Desmosterolosis, 602398
DHCR7	176.8	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHFR	55	90%	73%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	162.9	99%	97%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIAPH1	134.8	99%	97%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	188.7	99%	98%	Mental retardation, FRA12A type, 136630
DKC1	138.1	99%	98%	Dyskeratosis congenita, X-linked, 305000
DLD	142	99%	97%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	113	99%	95%	Mental retardation, X-linked 90, 300850
DMD	147.3	99%	97%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	127.2	99%	97%	Myotonic dystrophy 1, 160900
DNAJC19	105.3	97%	90%	3-methylglutaconic aciduria, type V, 610198
DNM1	171.5	94%	91%	Epileptic encephalopathy, early infantile, 31, 616346
DNMT3A	126	98%	95%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	141	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860

DOCK6	133.6	99%	97%	Adams-Oliver syndrome 2, 614219
DOCK7	128.8	97%	94%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	134.5	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	179.7	99%	99%	Developmental delay with short stature,dysmorphic features and sparse hair,616901
DPM1	136.9	89%	84%	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	177.9	95%	94%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DST	179.4	99%	98%	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653
DYM	118.2	96%	94%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	196.8	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	176.5	100%	99%	Mental retardation, autosomal dominant 7, 614104
EBP	101.3	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EDC3	153.4	100%	99%	?Mental retardation, autosomal recessive 50, 616460
EEF1A2	208.6	99%	98%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFTUD2	125.4	99%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	161.2	98%	96%	Kleefstra syndrome, 610253
EIF2AK3	169.7	96%	91%	Wolcott-Rallison syndrome, 226980
EIF4A3	122.1	100%	99%	Robin sequence with cleft mandible and limb abnormalities,268305
EIF4G1	144.4	99%	99%	{Parkinson disease 18}, 614251
ELOVL4	104.8	99%	97%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
EMC1	134.6	100%	99%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMX2	121.8	100%	99%	Schizencephaly, 269160
EP300	205.7	99%	98%	Colorectal cancer, somatic, 114500

				Rubinstein-Taybi syndrome 2, 613684
EPB41L1	139.7	100%	99%	?Mental retardation, autosomal dominant 11, 614257
ERCC2	143.5	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	117.6	99%	98%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC5	161.6	99%	99%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	192.3	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	95.1	93%	81%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	167.7	99%	99%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	121.1	95%	90%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	128.1	100%	100%	Glutaric acidemia IIB, 231680
ETHE1	86.4	99%	94%	Ethylmalonic encephalopathy, 602473
EXOSC3	81.7	95%	88%	Pontocerebellar hypoplasia, type 1B, 614678
EZH2	153.3	99%	98%	Weaver syndrome, 277590
FAM126A	144.4	97%	94%	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	91.8	96%	91%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FAT4	245.8	100%	99%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBN1	185.2	99%	99%	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185

				Marfan lipodystrophy syndrome,616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBXL4	227.1	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	111.7	95%	89%	?Mental retardation, autosomal recessive 45, 615979
FGD1	98.1	95%	89%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGFR1	161.5	99%	97%	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	156.4	96%	95%	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	120.2	99%	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	175.4	92%	88%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	93	99%	98%	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	157.4	98%	93%	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	161.1	99%	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	149.3	99%	96%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FMN2	97.2	84%	78%	Mental retardation, autosomal recessive 47, 616193
FMR1	93.9	95%	85%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360

FOXG1	130.8	89%	82%	Rett syndrome, congenital variant, 613454
FOXP1	148.5	100%	99%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	174.1	98%	95%	Speech-language disorder-1, 602081
FRAS1	165	100%	99%	Fraser syndrome, 219000
FREM2	199.1	99%	99%	Fraser syndrome, 219000
FRRS1L	143	69%	63%	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	99.6	94%	88%	Glutamate formiminotransferase deficiency, 229100
FTO	158.3	99%	99%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	140.6	99%	96%	Mental retardation, X-linked 9, 309549
FUCA1	156.2	99%	99%	Fucosidosis, 230000
GABRA1	199.9	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GAD1	132.5	99%	97%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALE	168.6	100%	100%	Galactose epimerase deficiency, 230350
GALT	168	100%	100%	Galactosemia, 230400
GAMT	119.1	97%	91%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	144.7	100%	99%	Mental retardation, autosomal dominant 18, 615074
GATM	174.8	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	91.2	95%	86%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	38.8	83%	62%	Glycine encephalopathy, 605899
GDI1	177.3	100%	100%	Mental retardation, X-linked 41, 300849
GFAP	110.3	99%	98%	Alexander disease, 203450
GJB1	237.6	100%	99%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	52.2	85%	68%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	51.7	77%	62%	Glycerol kinase deficiency, 307030
GLB1	93.9	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010

GLDC	90.1	91%	84%	Glycine encephalopathy, 605899
GLI2	139.5	98%	96%	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	170.9	100%	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	239	99%	99%	D-glyceric aciduria, 220120
GM2A	143.6	100%	99%	GM2-gangliosidosis, AB variant, 272750
GMPPA	148.3	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	256.1	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	204.8	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GNAS	140.1	98%	96%	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	212.5	100%	100%	Leukemia,acute lymphoblastic,somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNPAT	159.6	98%	95%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	192	98%	97%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNS	123	96%	91%	Mucopolysaccharidosis type IIID, 252940
GPC3	121	98%	94%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	192.1	98%	97%	Molybdenum cofactor deficiency C, 615501
GPR56	173.8	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752

GPT2	158.4	98%	93%	?Mental retardation, autosomal recessive 49, 616281
GRIA3	112.8	99%	95%	Mental retardation, X-linked 94, 300699
GRID2	195.3	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	153.9	95%	93%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	166.9	100%	99%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	176.5	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	213.2	99%	99%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIP1	152.3	99%	99%	Fraser syndrome, 219000
GRM1	194.5	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSS	113.1	100%	99%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	149.4	100%	99%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP3	135.8	99%	98%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	125.1	90%	87%	Mucopolysaccharidosis VII, 253220
HACE1	149.9	98%	94%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HAX1	148	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	123.6	100%	98%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	121.5	99%	97%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	142.6	99%	98%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	119.7	99%	99%	No OMIM phenotype
HDAC6	127	99%	97%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	165.1	100%	99%	Cornelia de Lange syndrome 5, 300882
HEPACAM	156.7	86%	78%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	199	99%	99%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	117.3	79%	75%	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	78.1	96%	87%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230

HEXA	143.7	100%	99%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	152.6	97%	91%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HIVEP2	210.1	100%	100%	Mental retardation, autosomal dominant 43, 616977
HLCS	193.1	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNMT	151.6	99%	98%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HOXA1	173.6	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HPD	151.2	100%	99%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	75.3	94%	84%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HRAS	195.3	99%	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	120.3	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSPD1	92.7	96%	89%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HUWE1	116.8	99%	97%	Mental retardation, X-linked syndromic, Turner type, 300706
IARS	171.3	99%	98%	No OMIM phenotype
IDS	119.1	99%	98%	Mucopolysaccharidosis II, 309900
IDUA	116.9	91%	85%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	60.9	86%	78%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	130	98%	96%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250

IFT172	128.2	99%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IGBP1	122.3	99%	96%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	146.8	100%	99%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	56.2	84%	70%	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	143.8	99%	97%	Mental retardation, X-linked 21/34, 300143
INPP5E	105.1	96%	91%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
IQSEC2	72.2	94%	86%	Mental retardation, X-linked 1/78, 309530
ISPD	120	95%	85%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITPR1	176.7	100%	99%	Gillespie syndrome,206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	126	99%	99%	Isovaleric acidemia, 243500
JAG1	167	99%	98%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAM3	166.6	99%	98%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANK1	179.6	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	94.7	94%	89%	Koolen-De Vries syndrome, 610443
KAT6A	199.3	100%	99%	Mental retardation, autosomal dominant 32, 616268
KAT6B	202.7	99%	98%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	156.4	100%	100%	Lissencephaly 6, with microcephaly, 616212
KCNA2	180.7	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	149.4	100%	99%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC3	155.3	71%	58%	Spinocerebellar ataxia 13, 605259
KCNH1	198.9	100%	99%	Temple-Baraitser syndrome, 611816

				Zimmermann-Laband syndrome 1, 135500
KCNJ10	229	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	302.7	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	196.9	100%	99%	Keppen-Lubinsky syndrome, 614098
KCNK9	195.6	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	103.9	98%	96%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNT1	129.2	95%	93%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	144.9	93%	92%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	154.5	98%	96%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5C	128.3	98%	95%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	127.5	93%	86%	Kabuki syndrome 2, 300867
KIAA0226	116.4	98%	97%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA1033	105.1	93%	88%	?Mental retardation, autosomal recessive 43, 615817
KIAA1279	198.7	100%	99%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	183	100%	99%	Mental retardation, X-linked 98, 300912
KIF11	91.9	96%	93%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	134.8	99%	97%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF2A	114.4	96%	88%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	103.8	94%	90%	?Mental retardation, X-linked 100, 300923
KIF5C	126.7	99%	98%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	93.4	95%	88%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131

				?Hydrolethalus syndrome 2, 614120
KIRREL3	160.2	99%	99%	Mental retardation, autosomal dominant 4, 612581
KMT2A	173.8	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage Wiedemann-Steiner syndrome, 605130
KMT2D	162.1	99%	99%	Kabuki syndrome 1, 147920
KPTN	118.2	100%	99%	Mental retardation, autosomal recessive 41, 615637
KRAS	72.1	99%	96%	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
KRBOX4	193.5	99%	99%	No OMIM phenotype nonsyndromic X-linked mental retardation (Lugtenberg et al. 2006)
L1CAM	157.7	99%	98%	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	139.8	98%	96%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	156.8	99%	99%	Poretti-Boltshauser syndrome, 615960
LAMA2	164.8	99%	99%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMC3	135.5	98%	95%	Cortical malformations, occipital, 614115
LAMP2	134.3	92%	91%	Danon disease, 300257
LARGE	145.9	99%	98%	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	61.2	77%	66%	Alazami syndrome, 615071
LIAS	159.6	99%	95%	Hyperglycinemia, lactic acidosis, and seizures, 614462

LIG4	181.8	100%	99%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LINS	157.3	99%	97%	Mental retardation, autosomal recessive 27, 614340
LMAN2L	143.9	99%	99%	?Mental retardation, autosomal recessive, 52,616887
LONP1	168.1	97%	95%	CODAS syndrome, 600373
LRP2	199.9	100%	99%	Donnai-Barrow syndrome, 222448
LRPPRC	140.1	98%	96%	Leigh syndrome, French-Canadian type, 220111
LZTFL1	136.5	98%	94%	Bardet-Biedl syndrome 17, 615994
MAGEL2	153.2	100%	99%	Schaaf-Yang syndrome, 615547
MAGT1	130.4	98%	95%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAN1B1	156.2	99%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	137.2	98%	96%	Mannosidosis, alpha-, types I and II, 248500
MANBA	144.2	99%	95%	Mannosidosis, beta, 248510
MAOA	142.8	100%	99%	Brunner syndrome, 300615 {Antisocial behavior},300615
MAP2K1	107.3	99%	95%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	118.5	97%	92%	Cardiofaciocutaneous syndrome 4, 615280
MAPRE2	226.5	99%	99%	Symmetric circumferential skin creases, congenital, 2, 616734
MAT1A	196.4	99%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	202.7	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	142.6	99%	97%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MCCC1	169.5	99%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	142	99%	98%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	166.6	98%	96%	Mucolipidosis IV, 252650
MCPH1	155.4	99%	97%	Microcephaly 1, primary, autosomal recessive, 251200
MECP2	100	99%	94%	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
MED12	116.5	98%	95%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	151.7	100%	99%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	148.5	99%	96%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	146.7	98%	96%	Mental retardation, autosomal recessive 18, 614249
MEF2C	142.4	98%	93%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
METTL23	150.3	100%	100%	Mental retardation, autosomal recessive 44, 615942
MGAT2	168.8	100%	99%	Congenital disorder of glycosylation, type IIa, 212066
MICU1	140	95%	91%	Myopathy with extrapyramidal signs, 615673
MID1	196.6	99%	98%	Opitz GBBB syndrome, type I, 300000
MID2	163.1	99%	98%	?Mental retardation, X-linked 101, 300928
MKKS	239.6	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MLC1	109.9	100%	99%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	93.2	94%	91%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	201.9	99%	99%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMACHC	205.3	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	82.8	87%	74%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	92.2	98%	93%	Molybdenum cofactor deficiency A, 252150
MOCS2	156.7	99%	98%	Molybdenum cofactor deficiency B, 252160
MOGS	121.3	99%	98%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	131.9	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPDZ	176.7	98%	96%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPLKIP	87.3	94%	81%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRPL3	69.9	89%	78%	Combined oxidative phosphorylation deficiency 9, 614582
MRPS22	150.8	95%	91%	Combined oxidative phosphorylation deficiency 5, 611719
MTHFR	153.2	100%	99%	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	152.8	100%	99%	Smith-Kingsmore syndrome, 616638
MTR	158.9	99%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	143	99%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	130.1	99%	95%	Methylmalonic aciduria, mut(0) type, 251000
MVK	146.1	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYCN	102.9	94%	85%	Feingold syndrome, 164280
MYH9	146.4	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	142.9	99%	97%	Griscelli syndrome, type 1, 214450
MYT1L	195.7	100%	99%	Mental retardation, autosomal dominant 39, 616521
NAA10	111	99%	96%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	162.6	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	123.7	93%	91%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NALCN	155.5	99%	97%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	123.8	100%	99%	Sponyloepimetaphyseal dysplasia, Genevieve type,610442
NBN	89.5	98%	94%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260

NDE1	105	100%	99%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDP	140	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDST1	220.2	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	236.8	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFA11	95.1	99%	94%	Mitochondrial complex I deficiency, 252010
NDUFA12	166	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFAF5	104.7	97%	94%	Mitochondrial complex 1 deficiency, 252010
NDUFS1	154.7	99%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	120.4	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	151.1	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	175.1	100%	98%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS7	132.1	99%	99%	Leigh syndrome, 256000
NDUFS8	145.6	99%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	168.6	99%	97%	Mitochondrial complex I deficiency, 252010
NECAP1	133.2	100%	100%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	163.6	99%	97%	No OMIM phenotype {Essential hypertension, association with} (Russo (2005) Hypertension 46,488) Epilepsy,photosensitive generalised (Dibbens (2007),Genes Brain Behav 6,750) Infantile spasms (Allen (2013) Nature 501,217) Impaired ENaC regulation (Fouladkou (2004) Am J Physiol Renal Physiol 287,F550)
NEU1	19.8	73%	44%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NF1	140.5	93%	89%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFIA	144.4	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	98%	95%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NHS	143.7	95%	93%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIPBL	129.3	96%	94%	Cornelia de Lange syndrome 1, 122470
NKX2-1	55.6	98%	93%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550
NLGN3	153.2	100%	99%	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	191	99%	98%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRP3	153.2	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NONO	111.4	99%	96%	Mental retardation, X-linked, syndromic 34, 300967
NPHP1	141	99%	97%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NR2F1	216.8	99%	98%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRAS	205.7	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	182	99%	97%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	172.3	100%	99%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626

				Sotos syndrome 1, 117550
NSDHL	205.2	99%	99%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	128.9	95%	93%	Mental retardation, autosomal recessive 5, 611091
NTRK1	142.6	99%	97%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUP62	126.9	99%	99%	Striatonigral degeneration, infantile, 271930
OCLN	215.3	100%	100%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	152.2	99%	97%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	56.1	84%	71%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	113.4	99%	96%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	125	99%	97%	Meier-Gorlin syndrome 1, 224690
OTC	143.9	100%	99%	Ornithine transcarbamylase deficiency, 311250
PACS1	130.6	97%	95%	Schuss-Hoeijmakers-syndrome, 615009
PAFAH1B1	116.2	89%	82%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	191.3	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	95.9	97%	91%	Mental retardation, X-linked 30/47, 300558
PANK2	177.5	99%	96%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX1	126.6	87%	81%	?Otofaciocervical syndrome 2, 615560
PAX6	156.1	100%	99%	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	104.2	100%	99%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PC	162.8	99%	97%	Pyruvate carboxylase deficiency, 266150
PCDH19	226.4	100%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	127.8	98%	95%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	126.3	98%	95%	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDHA1	127.8	97%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDSS1	134.8	91%	85%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	131.3	97%	93%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	121.4	99%	98%	Prolidase deficiency, 170100
PET100	127.6	95%	82%	Mitochondrial complex IV deficiency, 220110
PEX1	123.4	97%	95%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	118.3	97%	93%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	123.3	99%	99%	Peroxisome biogenesis disorder 14B, 614920
PEX12	165.4	100%	99%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	212.2	99%	98%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	138.5	96%	92%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	121	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	169.7	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	86.5	99%	99%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.8	98%	94%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	126.7	99%	97%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	92	90%	84%	Heimler syndrome 2, 616617

				Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	138.5	89%	85%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGAP1	109.7	93%	86%	Mental retardation, autosomal recessive 42, 615802
PGAP2	186.1	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	135.7	98%	95%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	60	92%	81%	Phosphoglycerate kinase 1 deficiency, 300653
PHF6	77.9	92%	83%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	110.5	99%	97%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	138.6	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	102.1	92%	84%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGG	196.9	99%	99%	Mental retardation,autosomal recessive 53,616917
PIGL	136.6	100%	98%	CHIME syndrome, 280000
PIGN	128.6	95%	89%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	140.6	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	174.6	99%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	171.1	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGY	113.6	100%	99%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3R2	96.5	89%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	132.4	99%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	177.3	99%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	162.1	100%	99%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLXND1	135	96%	93%	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	178.4	99%	99%	Congenital disorder of glycosylation, type Ia, 212065

PNKP	98.4	99%	97%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	148.5	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POC1A	150.5	100%	99%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POGZ	188.2	99%	98%	White-Sutton syndrome, 616364
POLG	126.2	99%	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	162.2	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	168.9	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	131.9	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	281.8	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	225.3	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	181	99%	97%	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	120.5	98%	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	145.4	100%	99%	Focal dermal hypoplasia, 305600
POU1F1	125.8	97%	93%	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	38.6	75%	62%	No OMIM phenotype ?Intellectual disability (Dheedene (2014) Mol Syndromol 5,32)
PPOX	108.1	99%	97%	Porphyria variegata, 176200
PPP2R1A	151.6	93%	92%	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	163.4	99%	99%	Mental retardation, autosomal dominant 35, 616355
PPT1	190.4	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730

PQBP1	174.3	97%	96%	Renpenning syndrome, 309500
PRODH	95.8	88%	82%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	201.5	100%	100%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRSS12	167.7	99%	98%	Mental retardation, autosomal recessive 1, 249500
PSAP	126.7	99%	98%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSEN1	161.6	99%	98%	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	138.6	98%	96%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCHD1	176.6	100%	99%	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	151.7	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	152.4	99%	98%	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	101.2	96%	90%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTS	128.2	97%	89%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	182.7	99%	98%	Verheij syndrome, 615583
PURA	125.2	98%	95%	Mental retardation, autosomal dominant 31, 616158
PUS1	150.8	99%	96%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	231.8	100%	100%	?Mental retardation, autosomal recessive 55, 617051
PUS7	174.8	99%	97%	No OMIM phenotype
PYCR1	105.4	99%	94%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	137.6	99%	98%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	167.3	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	88.5	100%	99%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	102.5	95%	85%	Warburg micro syndrome 3, 614222
RAB27A	178	100%	98%	Griscelli syndrome, type 2, 607624
RAB39B	139.4	100%	99%	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RAB3GAP1	143.3	99%	98%	Warburg micro syndrome 1, 600118
RAB3GAP2	104.5	97%	93%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	165.7	100%	100%	No OMIM phenotype
RAD21	97.7	99%	96%	Cornelia de Lange syndrome 4, 614701
RAF1	138	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAI1	142.2	100%	99%	Smith-Magenis syndrome, 182290
RARS2	126.3	99%	98%	Pontocerebellar hypoplasia, type 6, 611523
RBM10	125.8	99%	98%	TARP syndrome, 311900
RBM28	160.7	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	86.4	93%	85%	Adams-Oliver syndrome 3, 614814

RELN	181.5	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
RERE	83.2	96%	92%	Neurodevelopmental disorder with or without anomalies of the brain,eye or heart, 616975
REV3L	156.3	98%	96%	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	118.8	99%	97%	Congenital disorder of glycosylation, type In, 612015
RIT1	184.4	100%	100%	Noonan syndrome 8, 615355
RLIM	140.9	99%	98%	Mental Retardation, X-linked 61, 300978
RMND1	142.9	99%	96%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	149.3	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	125.1	94%	84%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	207.6	99%	97%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	89.1	91%	84%	Leukoencephalopathy, cystic, without megalencephaly, 612951
ROGDI	134	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPGRIP1L	153.6	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	88.9	98%	92%	{Autism, susceptibility to, X-linked 5}, 300847
RPS6KA3	99.5	94%	87%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RTEL1	127.4	99%	96%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTTN	152.5	97%	95%	Microcephaly,short stature,and polymicrogyria with seizures,614833
SALL1	147.9	99%	98%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SATB2	120.1	98%	93%	Glass syndrome, 612313
SBDS	210.4	99%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135

SC5D	227.6	99%	98%	Lathosterolosis, 607330
SCN1A	157.3	99%	98%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN2A	170.4	99%	97%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN8A	224.3	99%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SCO2	113.3	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	117.4	84%	78%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SEPSECS	185	99%	99%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	125.5	98%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	149.3	97%	96%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	124.5	98%	96%	No OMIM phenotype Schizophrenia (Takata (2014) Neuron 82, 723)
SETD2	170.9	99%	99%	Luscan-Lumish syndrome, 616831
SETD5	203.5	99%	99%	Mental retardation, autosomal dominant 23, 615761
SGSH	146.8	97%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SHANK2	147	99%	99%	{Autism susceptibility 17}, 613436
SHANK3	82.5	83%	73%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	110.4	98%	94%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	148.9	99%	98%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	115.8	99%	98%	?Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	99.2	98%	94%	Epileptic encephalopathy, early infantile, 30, 616341

SIL1	173.8	99%	98%	Marinesco-Sjogren syndrome, 248800
SIN3A	171.2	99%	98%	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
SIX3	157.3	99%	96%	Holoprosencephaly-2, 157170 Schizencephaly, 269160
SKI	90.1	98%	95%	Shprintzen-Goldberg syndrome, 182212
SLC12A6	169.6	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	176.9	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	69.6	96%	86%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	123.8	97%	93%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	191.3	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	221.3	100%	99%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A4	176.8	99%	97%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A12	165	99%	98%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	228.7	98%	95%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	117.1	99%	96%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	183.5	100%	100%	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	148.5	96%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A2	118.7	99%	96%	Congenital disorder of glycosylation, type IIm, 300896
SLC35C1	230	99%	97%	Congenital disorder of glycosylation, type IIc, 266265
SLC39A8	145.1	100%	99%	Congenital disorder of glycosylation, type IIn, 616721
SLC4A4	150.3	99%	98%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A17	220.5	100%	99%	Mental retardation, autosomal recessive 48, 616269
SLC6A3	153.5	100%	99%	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	61.1	92%	82%	Cerebral creatine deficiency syndrome 1, 300352

SLC7A7	127.2	100%	100%	Lysinuric protein intolerance, 222700
SLC9A6	126.9	97%	90%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	132.5	99%	98%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMARCA2	131.1	96%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	156.7	99%	98%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	248.9	100%	100%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	86.7	96%	88%	Coffin-Siris syndrome 5,616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	120	100%	99%	Cornelia de Lange syndrome 2,300590
SMC3	95.1	93%	86%	Cornelia de Lange syndrome 3, 610759
SMOC1	132.2	99%	97%	Microphthalmia with limb anomalies, 206920
SMPD1	134.3	99%	97%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	70.1	88%	76%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP29	160.1	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	156.6	99%	96%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRPN	130.4	99%	93%	Prader-Willi syndrome, 176270
SNX14	76.2	92%	82%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	119.1	95%	88%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	168.2	98%	94%	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179) ?Developmental delay,seizure disorder,macrocephaly and white matter abnormalities (Zhu (2015) Genet Med)
SOS1	106.9	96%	90%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOX10	80.4	97%	93%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584

				Waardenburg syndrome, type 4C, 613266
SOX11	125.8	99%	97%	Mental retardation, autosomal dominant, 27, 615866
SOX2	111.4	99%	97%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	45.5	94%	80%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	120	99%	97%	Lamb-Shaffer syndrome, 616803
SPATA5	146.6	99%	99%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG11	146.9	98%	96%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPRED1	187.2	98%	96%	Legius syndrome, 611431
SPTAN1	139.4	99%	98%	Epileptic encephalopathy, early infantile, 5, 613477
SPTBN2	122.1	99%	99%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SRCAP	162.7	99%	98%	Floating-Harbor syndrome, 136140
SRD5A3	166.9	99%	99%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	90.5	99%	98%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	193.7	100%	100%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	138.4	95%	94%	Amish infantile epilepsy syndrome, 609056
STAMBP	130.1	99%	96%	Microcephaly-capillary malformation syndrome, 614261
STIL	172.5	99%	98%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	131	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STT3A	192.3	100%	100%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	137.9	99%	96%	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	185.6	100%	99%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	147.9	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	69.4	92%	82%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	111.3	99%	97%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400

SUOX	219.5	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	97	89%	88%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYN1	79.6	83%	70%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	156.8	99%	99%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNGAP1	75	95%	86%	Mental retardation, autosomal dominant 5, 612621
SYP	77.8	99%	95%	Mental retardation, X-linked 96, 300802
SYT14	189.8	92%	84%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	157.5	99%	99%	Epileptic encephalopathy, early infantile, 18, 615476
TAF1	141.1	99%	97%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF2	136.8	98%	94%	Mental retardation, autosomal recessive 40, 615599
TAT	141.9	100%	100%	Tyrosinemia, type II, 276600
TBC1D20	163.7	94%	94%	Warburg micro syndrome 4, 615663
TBC1D24	178.8	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	118.1	97%	94%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	151.6	99%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	97.6	95%	89%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3,616900
TBL1XR1	97.3	90%	75%	Mental retardation, autosomal dominant 41,616944 Piermont syndrome,602342
TBR1	116.8	99%	97%	No OMIM phenotype Intellectual disability (Hamdan (2014) PLoS Genet 10) ?Autism (O'Roak (2012) Science 338,1619) ?Ventriculomegaly (Traylor (2012) Mol Syndromol 3,102)
TCF4	160.1	99%	99%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCTN3	135.8	99%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860

TECR	96.4	99%	97%	Mental retardation, autosomal recessive 14, 614020
TELO2	111.5	98%	93%	You-Hoover-Fong syndrome, 616954
TFAP2A	138	100%	99%	Branchiooculofacial syndrome, 113620
TGFBR1	213.8	95%	93%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	215.3	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	151.6	99%	99%	Holoprosencephaly-4, 142946
TH	83.6	97%	92%	Segawa syndrome, recessive, 605407
THOC2	92.7	95%	87%	Mental retardation, X-linked 12/35, 300957
THOC6	262	100%	99%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	189.5	99%	99%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	45.5	87%	70%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TINF2	196.8	100%	99%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMCO1	105.1	99%	98%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM165	122.2	98%	96%	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	103.4	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	115.6	99%	96%	Joubert syndrome 14, 614424
TMEM240	130.5	100%	99%	Spinocerebellar ataxia 21, 607454
TMEM67	78.9	92%	83%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	152.6	95%	91%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	116.6	99%	96%	{Autism,susceptibility to,X-linked 6}, 300872
TNIK	128.4	99%	98%	Mental retardation, autosomal recessive 54, 617028
TPP1	158.7	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500

				Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAPPC11	140.7	98%	95%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC9	154	100%	99%	Mental retardation, autosomal recessive 13, 613192
TREX1	272.2	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	152.9	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIO	154.1	98%	96%	Mental retardation, autosomal dominant 44,617061
TRMT10A	157.2	99%	98%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TSC1	149.6	99%	98%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	144.5	99%	98%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN54	94.9	94%	90%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSPAN7	145.4	99%	99%	Mental retardation, X-linked 58, 300210
TTC19	106.2	90%	81%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	139	99%	97%	Trichohepatoenteric syndrome 1, 222470
TTC8	109.1	98%	93%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	114.2	99%	98%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	117.1	99%	97%	Lissencephaly 3, 611603
TUBA8	178.3	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB	21.4	78%	50%	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB2B	109.8	100%	100%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	208.4	99%	98%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	137	96%	95%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	185.9	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412

TUBGCP4	158.9	99%	96%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	162.6	100%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	153.5	99%	98%	Mental retardation, autosomal recessive 7, 611093
TWIST1	143	95%	86%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Saethre-Chotzen syndrome, 101400
UBE2A	115.3	99%	95%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	103.9	98%	93%	Angelman syndrome, 105830
UBE3B	145.8	100%	99%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	144.4	98%	95%	Johanson-Blizzard syndrome, 243800
UNC80	146.1	99%	98%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	177.2	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	58.2	92%	80%	Mental retardation, X-linked, syndromic 14, 300676
UQCRQ	162.5	100%	99%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	150	100%	99%	?Urocanase deficiency, 276880
USP9X	142.5	97%	92%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
VLDLR	244.5	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13B	156.8	98%	96%	Cohen syndrome, 216550
VPS37A	79.7	86%	69%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	153.1	91%	90%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	147	98%	95%	Pontocerebellar hypoplasia type 1A, 607596
WAC	186.7	99%	96%	Desanto-Shinawi syndrome, 616708
WDR19	153.5	99%	97%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR45	92.9	97%	92%	Neurodegeneration with brain iron acculation 5, 300894
WDR62	170.9	99%	98%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	151.1	100%	99%	Galloway-Mowat syndrome, 251300
WDR81	171.3	99%	99%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	148	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239

				Spinocrebellar ataxia, autosomal recessive 12, 614322
XPA	58.3	95%	84%	Xeroderma pigmentosum, group A, 278700
XPNPEP3	148.8	98%	97%	Nephronophthisis-like nephropathy 1, 613159
XYLT1	148.3	93%	88%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	115.4	86%	79%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YWHAE	116.3	99%	96%	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)
ZBTB16	167.6	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	221.5	99%	98%	?Mental retardation, autosomal dominant 22, 612337
ZBTB20	248.6	100%	100%	Primrose syndrome, 259050
ZBTB24	191.3	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC3H14	213.6	99%	97%	No OMIM phenotype
ZDHHC15	104.9	98%	94%	?Mental retardation, X-linked 91, 300577
ZDHHC9	68.6	99%	92%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	181.5	100%	99%	Mowat-Wilson syndrome, 235730
ZFYVE26	134.1	99%	99%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	207	100%	100%	Craniosynostosis 6, 616602
ZIC2	111.1	89%	79%	Holoprosencephaly-5, 609637
ZMYND11	144.9	99%	99%	Mental retardation, autosomal dominant 30, 616083
ZNF41	136.4	100%	99%	Mental retardation, X-linked 89, 300848
ZNF592	140.6	100%	99%	Spinocerebellar ataxia, autosomal recessive 5, 251300
ZNF674	146.6	100%	100%	Mental retardation, X-linked 92, 300851
ZNF711	172.8	98%	96%	Mental retardation, X-linked 97, 300803
ZNF81	109.5	99%	96%	Mental retardation, X-linked 45, 300498
ZSWIM6	173.4	95%	92%	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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