INTELLECTUAL DISABILITY GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABCC9	107.1	100%	98%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	31.8	73%	64%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	100.4	100%	97%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	121.2	100%	100%	Chanarin-Dorfman syndrome, 275630
ACAD9	91.0	100%	99%	ACAD9 deficiency, 611126
ACO2	77.9	90%	83%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	73.9	99%	92%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	77.0	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	66.8	100%	97%	Mental retardation, X-linked 63, 300387
ACTB	60.1	97%	90%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	56.9	99%	93%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACVR1	91.8	100%	97%	Fibrodysplasia ossificans progressiva, 135100
ACY1	80.1	100%	97%	Aminoacylase 1 deficiency, 609924
ADAR	137.0	99%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADAT3	48.3	100%	99%	Mental retardation, autosomal recessive 36, 615286
ADCK3	97.8	100%	95%	Coenzyme Q10 deficiency, primary, 4, 612016
ADK	119.3	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	203.0	100%	100%	Mental retardation, autosomal dominant, 28, 615873
ADSL	127.3	100%	98%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AFF2	71.5	98%	95%	Mental retardation, X-linked, FRAXE type, 309548
AGA	117.2	100%	89%	Aspartylglucosaminuria, 208400
AGPAT2	60.2	94%	79%	Lipodystrophy, congenital generalized, type 1, 608594
AGTR2	103.5	100%	100%	Mental retardation, X-linked 88, 300852
AHCY	70.6	92%	73%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752

AHI1	115.0	100%	99%	Joubert syndrome-3, 608629
AIFM1	58.0	99%	87%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	125.6	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AK1	86.6	100%	98%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AKT3	115.6	100%	94%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALDH18A1	91.9	97%	91%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH3A2	97.1	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	70.5	94%	88%	Hyperprolinemia, type II, 239510
ALDH5A1	64.5	97%	94%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	47.7	45%	45%	ngenital disorder of glycosylation, type Ik, 608540
ALG12	103.4	100%	97%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	58.2	93%	85%	Congenital disorder of glycosylation, type Is, 300884
ALG2	115.0	100%	97%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	81.8	100%	93%	Congenital disorder of glycosylation, type Id, 601110
ALG6	104.1	100%	100%	Congenital disorder, type Ic, 603147
ALG9	83.5	100%	99%	Congenital disorder of glycosylation, type Il, 608776
ALX1	161.4	100%	100%	Frontonasal dysplasia 3, 613456
ALX4	80.4	100%	98%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMT	123.9	100%	99%	Glycine encephalopathy, 605899
ANK3	156.8	100%	99%	?Mental retardation, autosomal recessive, 37
ANKH	110.5	100%	99%	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKRD11	118.5	91%	87%	KBG syndrome, 148050
ANO10	102.8	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	83.3	97%	91%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S2	95.5	100%	100%	Mental retardation, X-linked syndromic, Fried type, 300630
AP3B1	109.5	100%	99%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	107.1	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	136.2	100%	99%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	102.6	100%	98%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	94.1	100%	100%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	121.3	96%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	114.4	100%	99%	Periventricular heterotopia with microcephaly, 608097

ARG1	131.7	98%	91%	Argininemia, 207800
ARHGEF6	54.8	97%	93%	Mental retardation, X-linked 46, 300436
ARHGEF9	50.9	98%	88%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	102.5	99%	95%	Mental retardation, autosomal dominant 14, 614607
ARID1B	122.4	100%	98%	Mental retardation, autosomal dominant 12, 614562
ARL13B	121.2	99%	95%	Joubert syndrome 8, 612291
ARL6	161.4	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARSE	48.8	91%	76%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	31.5	76%	62%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASL	80.4	99%	95%	Argininosuccinic aciduria, 207900
ASNS	70.1	91%	87%	Asparagine synthetase deficiency, 615574
ASPA	120.8	100%	99%	Canavan disease, 271900
ASPM	141.7	100%	99%	Microcephaly 5, primary, autosomal recessive, 608716
ASXL1	143.4	98%	96%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	157.3	100%	99%	Bainbridge-Ropers syndrome, 615485
ATIC	115.4	100%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A2	100.1	100%	98%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481
ATP2A2	120.5	100%	100%	Darier disease, 124200 Acrokeratosis verruciformis, 101900
ATP6AP2	31.7	93%	74%	Mental retardation, X-linked, with epilepsy, 300423
ATP6V0A2	106.5	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	60.3	100%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	95.3	98%	97%	?Cerebellar ataxia, mental retardation and dysequilibrium syndrome 4, 615268

ATR	119.4	100%	99%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	73.2	100%	98%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	114.5	100%	100%	3-methylglutaconic aciduria, type I, 250950
AUTS2	118.5	100%	100%	Mental Retardation, autosomal dominant 26, 615834
B3GALTL	107.6	95%	95%	Peters-plus syndrome, 261540
B4GALT1	78.9	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	89.7	100%	95%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
BBS1	114.8	100%	98%	Bardet-Biedl syndrome 1, 209900
BBS10	126.9	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	144.2	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	115.0	100%	99%	Bardet-Biedl syndrome 2, 209900
BBS4	97.2	97%	95%	Bardet-Biedl syndrome 4, 209900
BBS5	150.6	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	129.6	100%	99%	Bardet-Biedl syndrome 7, 209900
BBS9	124.3	100%	100%	Bardet-Biedl syndrome 9, 209900
BCKDHA	102.4	100%	98%	Maple syrup urine disease, type Ia, 248600
BCKDHB	85.2	98%	83%	Maple syrup urine disease, type Ib, 248600
BCOR	61.8	99%	95%	Microphthalmia, syndromic 2, 300166
BCS1L	140.9	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BLM	118.7	100%	100%	Bloom syndrome, 210900
BRAF	78.5	100%	99%	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707
BRWD3	62.9	98%	96%	Mental retardation, X-linked 93, 300659

BSCL2	110.4	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
BTD	141.7	100%	100%	Biotinidase deficiency, 253260
BUB1B	117.8	100%	98%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf57	77.6	100%	96%	Temtamy syndrome, 218340
C12orf65	174.3	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C5ORF42	129.8	100%	99%	Joubert syndrome 17, 614615
CA2	143.7	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA8	83.5	100%	100%	Cerebellar ataxia, mental retardation with/without quadrupedal locomotion 3, 613227
CACNG2	96.4	100%	100%	Mental retardation, autosomal dominant 10, 614256
CASK	51.4	98%	94%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CBL	127.5	100%	99%	onan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	77.6	99%	92%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	96.2	100%	97%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	93.6	100%	98%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCBE1	93.2	97%	89%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC78	90.8	100%	100%	Myopathy, centronuclear, 4, 614807
CDH15	80.2	100%	98%	Mental retardation, autosomal dominant 3, 612580
CDK5RAP2	110.5	99%	96%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	67.4	99%	96%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CDON	117.2	100%	98%	Holoprosencephaly 11, 614226
CENPJ	139.2	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	121.5	100%	99%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	133.3	99%	99%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823

CEP290	101.5	100%	98%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	84.5	100%	99%	Joubert syndrome 15, 614464
CHD2	128.0	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	121.0	100%	99%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	117.8	100%	100%	{Autism, susceptibility to, 18}, 615032
CHKB	91.1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CLCNKB	77.3	87%	81%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLIC2	33.1	92%	63%	Mental retardation, X-linked, syndromic 32, 300886
CLN3	85.1	100%	99%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.7	100%	90%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	63.7	98%	81%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	133.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNTNAP2	100.2	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COG1	121.0	100%	98%	Congenital disorder of glycosylation, type IIg, 611209
COG6	101.5	97%	95%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	77.0	100%	96%	Congenital disorder of glycosylation, type IIe, 608779
COG8	116.5	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	87.4	99%	96%	Porencephaly 1, 175780 Brain small vessel disease with hemorrhage, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773 Brain small vessel disease with Axenfeld-Rieger anomaly, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	80.1	99%	97%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COLEC11	121.8	100%	100%	3MC syndrome 2, 265050

COQ2	75.5	99%	96%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COX10	130.3	100%	97%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	73.4	100%	93%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
CPS1	107.7	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CRADD	128.0	100%	98%	Mental retardation, autosomal recessive 34, 614499
CRBN	145.0	100%	100%	Mental retardation, autosomal recessive 2, 607417
CREBBP	83.6	99%	97%	Rubinstein-Taybi syndrome, 180849
CTCF	114.3	100%	98%	Mental retardation, autosomal dominant 21, 615502
CTDP1	70.3	89%	87%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNB1	120.2	100%	99%	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Pilomatricoma, somatic, 132600 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTSA	96.1	100%	99%	Galactosialidosis, 256540
CTSD	96.2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	120.8	99%	96%	No OMIM phenotype Autism (Iossifov (2012) Neuron 74,285)
CUBN	88.4	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
CUL4B	61.7	100%	93%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CYB5R3	79.4	100%	96%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
D2HGDH	57.7	96%	86%	D-2-hydroxyglutaric aciduria, 600721
DARS2	116.2	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	111.7	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	97.1	99%	94%	Woodhouse-Sakati syndrome, 241080
DCX	60.6	100%	94%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDHD2	106.9	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033

DDX11	11.0	19%	14%	Warsaw breakage syndrome, 613398
DEAF1	68.5	83%	83%	Mental retardation, autosomal dominant 24, 615828
DHCR24	86.6	99%	97%	Desmosterolosis, 602398
DHCR7	115.9	100%	97%	Smith-Lemli-Opitz syndrome, 270400
DHFR	51.9	81%	58%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	103.3	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIP2B	100.8	100%	97%	Mental retardation, FRA12A type, 136630
DKC1	50.0	99%	91%	Dyskeratosis congenita, X-linked, 305000
DLD	143.7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	39.2	91%	79%	Mental retardation, X-linked 90, 300850
DMD	57.4	98%	95%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DMPK	96.7	100%	99%	Myotonic dystrophy 1, 160900
DNAJC19	57.8	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNMT3A	81.8	100%	99%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	91.6	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK8	83.2	100%	98%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DPAGT1	90.9	99%	95%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	157.7	100%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPYD	120.4	98%	96%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DST	170.6	100%	100%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DYM	102.2	100%	99%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326 Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DYNC1H1	117.4	99%	96%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	138.5	99%	99%	Mental retardation, autosomal dominant 7, 614104
EBP	43.9	94%	73%	Chondrodysplasia punctata, X-linked dominant, 302960
EFTUD2	89.7	98%	97%	Mandibulofacial dysostosis, Guion-Almeida type, 610536

EHMT1	100.4	98%	95%	Kleefstra syndrome, 610253
EIF2AK3	115.2	93%	92%	Wolcott-Rallison syndrome, 226980
EIF4G1	104.5	100%	99%	Parkinsons disease 18, 614251
ELOVL4	102.9	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMX2	105.1	100%	100%	Schizencephaly, 269160
EP300	139.2	99%	98%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPB41L1	86.4	98%	94%	Mental retardation, autosomal dominant 11, 614257
ERCC2	87.9	99%	96%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	123.3	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC5	122.5	99%	97%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	153.2	100%	98%	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980
ERCC8	86.0	100%	100%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	107.9	100%	98%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	96.3	100%	99%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	107.8	100%	99%	Glutaric acidemia 2B, 231680
ETHE1	61.0	100%	96%	Ethylmalonic encephalopathy, 602473
EXOSC3	64.3	94%	80%	Pontocerebellar hypoplasia, type 1B, 614678
FAM126A	143.6	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532

FBN1	98.5	100%	99%	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370 Geleophysic dysplasia 2, 614185
FGD1	47.0	94%	88%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGFR1	124.7	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465
FGFR2	116.5	100%	98%	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Scaphocephaly and Axenfeld-Rieger anomaly LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592

FGFR3	66.7	93%	90%	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500 Cervical cancer, somatic, 603956 LADD syndrome, 149730 CATSHL syndrome, 610474 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Spermatocytic seminoma, somatic, 273300
FH	85.7	98%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	80.2	100%	98%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 613153 Muscular dystrophy-dystroglycanopathy (with or without mental retardation), 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	108.0	100%	99%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 253800 Muscular dystrophy-dystroglycanopathy (without mental retardation), 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNA	62.1	99%	91%	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048
FLVCR1	95.8	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FMR1	55.2	98%	94%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360

FOXG1	91.1	93%	80%	Rett syndrome, congenital variant, 613454
FOXP1	94.4	99%	96%	Mental retardation with language impairment and autistic features, 613670
FOXP2	113.9	100%	100%	Speech-language disorder-1, 602081
FRAS1	98.6	98%	95%	Fraser syndrome, 219000
FTO	126.9	97%	97%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	52.4	92%	76%	Mental retardation, X-linked 9, 309549
FUCA1	74.3	100%	96%	Fucosidosis, 230000
GAD1	97.8	100%	100%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALE	108.6	100%	100%	Galactose epimerase deficiency, 230350
GALT	115.6	100%	100%	Galactosemia, 230400
GAMT	103.3	98%	94%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	112.9	100%	98%	Mental retardation, autosomal dominant 18, 615074
GATM	88.8	100%	94%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	109.0	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	14.8	52%	39%	Glycine encephalopathy, 605899
GDI1	70.5	99%	96%	Mental retardation, X-linked 41, 300849
GFAP	76.4	100%	97%	Alexander disease, 203450
GJB1	85.4	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	57.0	99%	83%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GK	24.7	77%	56%	Glycerol kinase deficiency, 307030
GLB1	73.6	99%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	60.1	98%	85%	Glycine encephalopathy, 605899
GLI2	107.0	100%	97%	Holoprosencephaly-9, 610829
GLI3	121.7	100%	99%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200
GM2A	107.6	100%	100%	GM2-gangliosidosis, AB variant, 272750

GMPPB	118.5	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A,14, 6135350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAO1	105.1	100%	99%	Epileptic encephalopathy, early infantile, 17, 615473
GNAS	118.1	100%	100%	Pseudohypoparathyroidism Ia, 103580 McCune-Albright syndrome, 174800 Pseudohypoparathyroidism Ic, 612462 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 Prolonged bleeding time, brachydactyly and mental retardation Acromegaly, 102200 Pseudopseudohypoparathyroidism, 612463 Prolonged bleeding time, brachydactyly, and mental retardation ACTH-independent macronodular adrenal hyperplasia, 219080
GNPAT	115.9	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNS	80.0	94%	85%	Mucopolysaccharidosis type IIID, 252940
GPC3	52.7	100%	97%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	120.0	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GPR56	94.6	100%	99%	Polymicrogyria, bilateral frontoparietal, 606854
GRIA3	56.7	91%	84%	Mental retardation, X-linked 94, 300699
GRIK2	119.5	100%	99%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	83.0	99%	93%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	132.6	99%	98%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	140.6	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRM1	152.4	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSE1	76.4	99%	92%	No OMIM phenotype Autism (Sanders (2012) Nature 485,237)
GSS	80.0	98%	94%	Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	97.0	100%	100%	Trichothiodystrophy, complementation group A, 601675
GUSB	61.9	89%	81%	Mucopolysaccharidosis VII, 253220
HAX1	140.7	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	63.9	100%	96%	Microphthalmia, syndromic 7, 309801
HCFC1	35.7	93%	78%	Mental retardation, X-linked 3, 309541

HDAC4	70.2	95%	88%	Brachydactyly-mental retardation syndrome, 600430
HDAC6	58.0	92%	84%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	48.3	99%	83%	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882
HERC2	62.9	63%	60%	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	99.0	100%	99%	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	90.7	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	114.8	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HLCS	141.8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HOXA1	141.0	100%	100%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HPD	98.3	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	50.8	100%	78%	Lesch-Nyhan syndrome, 300322
HRAS	94.7	100%	100%	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
HSD17B10	54.1	95%	91%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSPD1	14.8	61%	36%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HUWE1	48.2	96%	86%	Mental retardation, X-linked syndromic, Turner type, 300706
IDS	53.4	83%	75%	Mucopolysaccharidosis II, 309900
IDUA	86.0	95%	84%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015

IER3IP1	67.1	100%	99%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFT172	97.6	100%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IGBP1	49.0	84%	74%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	139.1	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	12.6	26%	24%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 {Atypical mycobacteriosis, familial}, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL1RAPL1	68.7	100%	99%	Mental retardation, X-linked 21/34, 300143
INPP5E	74.2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
IQSEC2	42.2	86%	69%	Mental retardation, X-linked 1, 309530
ISPD	88.9	95%	94%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
JAG1	110.2	99%	96%	Alagille syndrome, 118450 Deafness, congenital heart defects and posterior embryotoxon Tetralogy of Fallot, 187500
JAM3	71.1	94%	89%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANK1	137.3	100%	99%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	49.7	78%	65%	Koolen-De Vries syndrome, 610443
KAT6B	148.4	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNJ10	151.4	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	144.0	100%	100%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582
KCNK9	134.4	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292

KCNQ2	75.6	100%	98%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KCNT1	75.0	99%	93%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCTD7	106.3	92%	87%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	54.7	100%	93%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	64.4	99%	92%	Kabuki syndrome 2, 300867
KIAA0226	93.8	99%	96%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA1033	115.5	100%	98%	?Mental retardation, autosomal recessive 43, 615817
KIAA1109	126.9	100%	98%	No OMIM phenotype Schizophrenia (Gulsuner (2013) Cell 154,518)
KIAA1279	105.1	99%	95%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	78.3	100%	99%	Mental retardation, X-linked 98, 300912
KIF11	98.6	99%	96%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF5C	81.2	99%	94%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	67.0	93%	86%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	70.5	97%	91%	Mental retardation, autosomal dominant 4, 612581
KMT2A	142.0	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	100.9	99%	98%	Kabuki syndrome 1, 147920
KRAS	64.8	100%	87%	Lung cancer, somatic, 211980
L1CAM	62.6	98%	92%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100
L2HGDH	81.2	100%	97%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	97.2	99%	95%	Poretti-Boltshauser syndrome, 615960
LAMA2	102.7	100%	98%	Muscular dystrophy, congenital, 607855
LAMC3	95.8	99%	94%	Cortical malformations, occipital, 614115
LAMP2	62.5	97%	88%	Danon disease, 300257

LARGE	105.4	99%	95%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 613154 Muscular dystrophy-dystroglycanopathy (with mental retardation), 608840
LARP7	119.3	100%	100%	Alazami syndrome, 615071
LIG4	190.1	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LINS	124.2	100%	100%	Mental retardation, autosomal recessive 27, 614340
LRP2	107.4	100%	99%	Donnai-Barrow syndrome, 222448
LRPPRC	100.6	98%	96%	Leigh syndrome, French-Canadian type, 220111
MAGEL2	84.1	95%	91%	Prader-Willi-like syndrome, 615547
MAGT1	54.0	100%	100%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, magnesium defect, Epstein-Barr and neoplasia, 300853
MAN1B1	96.6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	80.2	97%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	92.3	100%	99%	Mannosidosis, beta, 248510
MAOA	55.6	100%	95%	Brunner syndrome, 300615
MAP2K1	103.7	95%	88%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	101.9	100%	98%	Cardiofaciocutaneous syndrome 4, 615280
MAT1A	94.5	100%	99%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	140.8	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	71.3	100%	98%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MCCC1	97.0	99%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	113.1	98%	92%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	96.7	97%	92%	Mucolipidosis IV, 252650
MCPH1	122.8	100%	100%	Microcephaly 1, primary, autosomal recessive, 251200
MECP2	90.5	99%	92%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 Angelman syndrome, 105830 Mental retardation, X-linked syndromic, Lubs type, 300260

MED12	62.1	94%	85%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MED13L	116.1	100%	99%	Transposition of the great arteries, dextro-looped 1, 608808
MED17	149.7	100%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	113.7	100%	100%	Mental retardation, autosomal recessive 18, 614249
MEF2C	113.4	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443
MGAT2	217.2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MID1	82.2	99%	93%	Opitz GBBB syndrome, type I, 300000
MKKS	148.8	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MLYCD	78.3	94%	81%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	174.5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMACHC	177.9	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MOCS1	73.9	100%	96%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	99.4	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MOGS	126.4	100%	100%	Congenital disorder of gycosylation, type 2b, 606056
MPDU1	113.2	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPDZ	103.8	98%	97%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPLKIP	91.9	100%	100%	Trichothiodystrophy, nonphotosensitive 1, 234050
MRPS22	90.7	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MTHFR	93.8	100%	98%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTR	104.3	100%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	105.2	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MUT	124.8	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MVK	88.3	100%	97%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYCN	107.3	97%	94%	Feingold syndrome, 164280

MYO5A	95.8	99%	97%	Griscelli syndrome, type 1, 214450
NAA10	51.9	97%	89%	N-terminal acetyltransferase deficiency, 300855
NAGA	78.6	100%	99%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	67.1	94%	89%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	109.7	99%	97%	?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419
NBN	121.4	98%	97%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NDE1	94.1	100%	95%	Lissencephaly 4 (with microcephaly), 614019
NDP	52.2	86%	78%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NDUFA1	116.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	103.4	99%	82%	Mitochondrial complex I deficiency, 252010
NDUFA12	96.2	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFS1	80.4	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	123.1	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS3	153.9	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	127.6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS7	100.0	100%	100%	Leigh syndrome, 256000
NDUFS8	107.7	100%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	63.4	100%	92%	Mitochondrial complex I deficiency, 252010
NEU1	13.5	64%	20%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NF1	82.7	84%	81%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Melanoma, desmoplastic neurotrophic (2) Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFIX	131.4	100%	98%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NHS	65.9	92%	88%	Nance-Horan syndrome, 302350

				Cataract 40, X-linked, 302200
NIPBL	122.3	98%	98%	Cornelia de Lange syndrome 1, 122470
NKX2-1	89.6	100%	100%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN3	61.6	99%	97%	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLRP3	124.1	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
NPHP1	169.1	100%	100%	hronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NR2F1	164.8	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRAS	129.6	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500
NRXN1	123.9	99%	98%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	123.9	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
NSDHL	48.0	96%	93%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	128.7	99%	93%	Mental retardation, autosomal recessive 5, 611091
NTRK1	67.6	98%	93%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
OCLN	95.8	72%	72%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	58.4	97%	94%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	37.0	88%	78%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804

OPHN1	48.4	99%	88%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	103.4	100%	99%	Meier-Gorlin syndrome 1, 224690
OTC	54.5	100%	94%	CGD Ornithine transcarbamylase deficiency, 311250
PACS1	104.1	98%	97%	Mental retardation, autosomal dominant 17, 615009
PAFAH1B1	71.2	89%	80%	Lissencephaly, 607432 Subcortical laminar heterotopia, 607432
PAH	86.5	100%	98%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	51.1	100%	97%	Mental retardation, X-linked 30/47, 300558
PANK2	112.1	90%	86%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX1	78.0	85%	66%	?Orofaciocervical syndrome 2, 615560
PAX6	83.9	100%	100%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700
PAX8	65.2	100%	86%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PC	99.2	95%	92%	Pyruvate carboxylase deficiency, 266150
PCDH19	67.3	100%	98%	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	94.7	98%	94%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDHA1	61.6	98%	89%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDSS1	94.4	90%	86%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	90.7	100%	99%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	66.9	97%	89%	Prolidase deficiency, 170100
PEX1	124.5	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73.0	95%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871

PEX11B	157.2	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	133.0	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	134.6	100%	95%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	98.5	94%	91%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	104.7	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	159.3	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	107.1	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	141.1	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	85.2	97%	95%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	91.3	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	102.2	99%	93%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGAP2	126.8	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	42.7	75%	67%	Phosphoglycerate kinase 1 deficiency, 300653
PHF6	71.5	100%	98%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	51.2	97%	85%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	84.6	100%	97%	Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	122.9	99%	99%	No OMIM phenotype Intellectual disability (de Ligt (2012) N Eng J Med 367,1921)
PIGL	95.7	100%	100%	CHIME syndrome, 280000
PIGN	106.6	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	104.6	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	133.6	100%	99%	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	182.7	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3R2	80.8	96%	87%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PLA2G6	76.5	99%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953

PLCB1	113.0	100%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	45.9	100%	94%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	92.4	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	70.4	100%	99%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	124.9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POC1A	103.3	96%	89%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POGZ	124.6	100%	99%	Autism (Neale (2012) Nature 485, 242) Intellectual disability (Gilissen (2014) Nature 511, 344) Schizophrenia (Fromer (2014) Nature 506, 179)
POLR3A	90.6	99%	96%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	104.2	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	99.1	100%	99%	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
POMT1	104.0	100%	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	74.5	100%	93%	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	58.7	92%	82%	Focal dermal hypoplasia, 305600
POU1F1	105.7	100%	100%	Pituitary hormone deficiency, combined, 1, 613038
PPOX	90.4	100%	97%	Porphyria variegata, 176200
PPT1	72.0	100%	96%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	68.1	100%	99%	Renpenning syndrome, 309500
PRODH	51.5	84%	66%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850

PRPS1	62.8	99%	97%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PRSS12	99.7	98%	96%	Mental retardation, autosomal recessive 1, 249500
PSAP	81.9	100%	98%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSEN1	96.5	100%	93%	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	83.9	99%	96%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTDSS1	117.4	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	137.7	100%	97%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Thyroid carcinoma, follicular, somatic, 188470 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355

PTPN11	46.9	88%	66%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PURA	111.3	100%	99%	Mental retardation, autosomal dominant 31, 616158
PUS1	65.8	100%	98%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	88.5	100%	96%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB18	111.7	100%	100%	Warburg micro syndrome 3, 614222
RAB27A	120.4	100%	100%	Griscelli syndrome, type 2, 607624
RAB39B	86.9	100%	100%	Mental retardation, X-linked 72, 300271
RAB3GAP1	124.9	99%	97%	Warburg micro syndrome 1, 600118
RAB3GAP2	110.7	99%	99%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	16.2	69%	39%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519
RAD21	97.2	99%	95%	Cornelia de Lange syndrome 4, 614701
RAF1	90.7	100%	100%	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAI1	137.2	99%	99%	Immunodeficiency 9, 612782 Smith-Magenis syndrome, 182290
RARS2	83.0	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RBM10	50.2	93%	83%	TARP syndrome, 311900
RBM28	103.7	100%	99%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RELN	105.8	99%	98%	Lissencephaly 2 (Norman-Roberts type), 257320
RFT1	78.3	100%	97%	Congenital disorder of glycosylation, type In, 612015
RIT1	162.5	100%	100%	Noonan syndrome 8, 615355
RMND1	86.7	95%	92%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH2A	90.2	100%	88%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	113.0	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	161.5	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	102.0	100%	98%	Leukoencephalopathy, cystic, without megalencephaly, 612951
ROGDI	94.4	96%	95%	Kohlschutter-Tonz syndrome, 226750
RPGRIP1L	101.3	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RPL10	47.0	97%	81%	{Autism, susceptibility to, X-linked 5}, 300847

RPS6KA3	50.9	98%	91%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RTEL1	81.0	99%	96%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
SALL1	145.0	100%	98%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SATB2	112.1	98%	96%	Cleft palate and mental retardation, 119540
SCN1A	118.1	99%	98%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403
SCN2A	128.1	100%	98%	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721
SCN8A	136.3	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCO2	85.4	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	9.1	30%	16%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SERAC1	89.8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	146.0	98%	96%	Schinzel-Giedion midface retraction syndrome, 269150
SETD5	155.1	100%	97%	No OMIM phenotype Autism (Neale (2012) Nature 485, 242) Intellectual disability (Grozeva (2014) Am J Hum Genet 94, 618)
SGSH	76.0	94%	94%	Mucopolysaccharidisis type 3A (Sanfilippo A), 252900
SHANK2	106.6	99%	96%	{Autism susceptibility 17}, 613436
SHANK3	66.6	93%	77%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	95.9	99%	85%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SHOC2	114.9	100%	99%	Noonan-like syndrome with loose anagen hair, 607721

SHROOM4	63.7	98%	94%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIL1	97.4	100%	95%	Marinesco-Sjogren syndrome, 248800
SIX3	119.8	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SKI	59.7	82%	79%	Shprintzen-Goldberg syndrome, 182212
SLC12A6	96.1	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	50.4	99%	92%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	100.9	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC1A1	122.3	100%	100%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC25A15	100.6	88%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	75.4	100%	92%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	94.3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC33A1	100.8	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A2	53.6	100%	98%	Congenital disorder of glycosylation, type 2m, 300896
SLC35C1	100.2	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC4A4	107.4	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A3	77.2	100%	100%	Parkinsonism -dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	4.4	13%	5%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	90.3	100%	100%	Lysinuric protein intolerance, 222700
SLC9A6	57.3	98%	90%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	134.5	100%	99%	Pancreatic cancer Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210
SMARCA2	90.1	97%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	83.0	97%	92%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMARCB1	126.2	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322

				Mental retardation, autosomal dominant 15, 614608
SMARCC2	88.3	97%	93%	No OMIM phenotype Autism (Neale (2012) Nature 485,242) Ivemark syndrome (Carss (2014) Hum Mol Genet 23, 3269)
SMC1A	70.7	97%	88%	Cornelia de Lange syndrome 2, 300590
SMC3	116.6	99%	98%	Cornelia de Lange syndrome 3, 610759
SMOC1	80.4	100%	96%	Microphthalmia with limb anomalies, 206920
SMPD1	109.7	99%	91%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	13.1	56%	30%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP29	128.5	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma , 609528
SNIP1	142.4	100%	99%	Psychomotor retardation, epilepsy and craniofacial dysmorphism, 614501
SOBP	118.7	100%	94%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOS1	122.2	100%	100%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SOX10	85.0	100%	100%	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136
SOX2	129.8	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	49.4	91%	84%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SPG11	110.5	100%	99%	Spastic paraplegia 11, autosomal recessive, 604360
SPRED1	135.9	100%	100%	Legius syndrome, 611431
SRCAP	134.9	100%	99%	Floating-Harbor syndrome, 136140
SRD5A3	130.2	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	46.7	97%	82%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	113.8	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	103.9	93%	93%	Amish infantile epilepsy syndrome, 609056
STAG1	96.9	100%	98%	No OMIM phenotype Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub) Intellectual disability (Gilissen (2014) Nature 511,344)
STIL	154.8	100%	100%	Microcephaly 7, primary, autosomal recessive, 612703

STRA6	68.9	100%	96%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STT3A	119.4	100%	99%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	104.4	100%	97%	?Congenital disorder of glycosylation, type Ix, 615597
STXBP1	93.2	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	81.6	94%	91%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUOX	170.4	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	91.8	88%	88%	Leigh syndrome, due to COX deficiency, 256000
SYN1	33.0	69%	52%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	53.6	92%	77%	Mental retardation, autosomal dominant 5, 612621
SYP	50.2	99%	92%	Mental retardation, X-linked 96, 300802
SYT14	127.4	93%	93%	Spinocerebellar ataxia, autosomal recessive 11, 614229
TAF2	106.2	100%	100%	Mental retardation, autosomal recessive 40, 615599
TAT	100.6	100%	100%	Tyrosinemia, type II, 276600
TBC1D24	106.0	100%	100%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TBC1D7	130.2	100%	100%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	118.9	100%	100%	Kenny-Caffey syndrome-1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410
TCF4	101.3	100%	100%	Pitt-Hopkins syndrome, 610954
TECR	81.1	100%	89%	Mental retardation, autosomal recessive 14, 614020
TFAP2A	72.6	95%	88%	Branchiooculofacial syndrome, 113620
TGFBR1	126.6	99%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	90.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
THOC6	156.2	100%	97%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	117.8	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	26.2	73%	66%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700

				Jensen syndrome, 311150
TMCO1	80.5	100%	100%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM165	92.6	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	73.6	97%	90%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	100.1	100%	94%	Joubert syndrome 14, 614424
TMEM67	119.5	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TMLHE	37.6	81%	74%	Epsilon-trimethyllysine hydrolylase deficiency, 300872
TPP1	135.3	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TRAPPC9	68.6	98%	91%	Mental retardation, autosomal recessive 13, 613192
TREX1	120.7	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	106.2	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110
TRIO	101.5	97%	93%	No OMIM phenotype Intellectual disability (de Ligt (2012) N Eng J Med 367,1921) Autism (Sanders (2012) Nature 485, 237)
TRMT10A	127.4	100%	100%	Microcephaly, short stature and impaired glucose metabolism, 616033
TSC1	93.2	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	84.0	99%	95%	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
TSEN54	105.5	98%	96%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSPAN7	43.6	95%	71%	Mental retardation, X-linked 58, 300210
TTC8	111.8	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464

TTI2	103.1	100%	100%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	19.9	84%	50%	Lissencephaly 3, 611603
TUBA8	100.5	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB2B	54.4	98%	92%	Polymicrogyria, symmetric or asymmetric, 610031
TUBGCP6	116.4	100%	98%	Microcephaly and chorioretinopathy, autosomal recessive 1, 251270
TUSC3	128.1	100%	100%	Mental retardation, autosomal recessive 7, 611093
TWIST1	99.1	100%	99%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400
UBE2A	57.6	100%	100%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	103.5	100%	100%	Angelman syndrome, 105830
UBE3B	108.2	99%	98%	Blepharophimosis-ptosis-intellectual disability syndrome, 615057
UBR1	104.7	100%	99%	Johanson-Blizzard syndrome, 243800
UPB1	110.4	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	55.9	98%	85%	Mental retardation, X-linked, syndromic 14, 300676
USP9X	69.6	99%	93%	Mental retardation, X-linked 99, 300919
VLDLR	117.9	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13B	114.0	99%	98%	Cohen syndrome, 216550
VRK1	133.6	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
WDR45	42.9	100%	89%	Neurodegeneration with brain iron acculation 5, 300894
WDR62	100.3	98%	94%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
XPA	87.6	100%	98%	Xeroderma pigmentosum, group A, 278700
XPNPEP3	137.5	100%	99%	Nephronophthisis-like nephropathy 1, 613159
XYLT1	106.0	94%	84%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YY1	85.2	100%	100%	No OMIM phenotype Mental retardation (Vissers (2010) Nat Genet 42, 1109)
ZBTB16	124.4	100%	98%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	158.2	99%	98%	?Mental retardation, autosomal dominant 22, 612337
ZDHHC15	60.5	100%	98%	?Mental retardation, X-linked 91, 300577
ZDHHC9	38.8	98%	88%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	155.1	100%	99%	Mowat-Wilson syndrome, 235730

ZFYVE26	88.6	96%	93%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC2	58.5	93%	86%	Holoprosencephaly-5, 609637
ZMYND11	119.5	100%	98%	Mental retardation, autosomal dominant 30, 616083
ZNF292	160.9	99%	98%	No OMIM phenotype Autism (Neale (2012) Nature 485,242) Potential protein deficiency (de Ligt (2012) N Engl J Med 367,1921)
ZNF41	64.1	100%	98%	Mental retardation, X-linked 89, 300848
ZNF592	114.5	94%	92%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF674	62.0	100%	87%	Mental retardation, X-linked 92, 300851
ZNF711	82.6	100%	100%	Mental retardation, X-linked 97, 300803
ZNF81	46.3	99%	96%	Mental retardation, X-linked 45, 300498

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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
OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors