INTELLECTUAL DISABILITY GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
A2ML1	132.9	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)
ABCC9	156.9	100%	99%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	49.6	69%	64%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	117.2	98%	93%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	244.7	100%	100%	Chanarin-Dorfman syndrome, 275630
ACAD9	137.4	99%	97%	ACAD9 deficiency, 611126
ACO2	111.4	95%	89%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	163.5	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	122.6	100%	97%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	70.3	92%	81%	Mental retardation, X-linked 63, 300387
ACTB	106.7	100%	93%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	110	100%	100%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACVR1	169	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ACY1	127.7	100%	97%	Aminoacylase 1 deficiency, 609924
ADAR	107.2	99%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADAT3	68.3	97%	84%	Mental retardation, autosomal recessive 36, 615286
ADCK3	114.9	99%	97%	Coenzyme Q10 deficiency, primary, 4, 612016
ADK	95.5	99%	95%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	236.3	100%	100%	Mental retardation, autosomal dominant, 28, 615873
ADSL	175.6	100%	100%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050

AFF2	87.3	100%	98%	Mental retardation, X-linked, FRAXE type, 309548
AGA	134	100%	100%	Aspartylglucosaminuria, 208400
AGO2	124.3	99%	99%	No OMIM phenotype Epithelial ovarian cancer,reduced risk,association with (Permuth-Wey (2011) Cancer Res 71,3896)
AGPAT2	101.5	100%	94%	Lipodystrophy, congenital generalized, type 1, 608594
AHCY	107.5	100%	99%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	103.2	98%	95%	Xia-Gibbs syndrome,615829
AHI1	133	99%	93%	Joubert syndrome-3, 608629
AIFM1	76.8	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	82.2	95%	86%	Leukodystrophy, hypomyelinating, 3, 260600
AK1	104	100%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AKT3	73.2	99%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALDH18A1	125.3	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH3A2	133.4	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	107.5	100%	99%	Hyperprolinemia, type II, 239510
ALDH5A1	76.6	84%	78%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	49.2	50%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALG12	145.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	57.3	97%	89%	Congenital disorder of glycosylation, type Is, 300884
ALG2	98.1	100%	98%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	101.1	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	91.8	95%	94%	Congenital disorder, type Ic, 603147
ALG9	118.5	98%	95%	Congenital disorder of glycosylation, type Il, 608776
ALX1	147.3	99%	95%	Frontonasal dysplasia 3, 613456
ALX4	112.6	97%	85%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMPD2	120.3	100%	99%	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	144.4	100%	100%	Glycine encephalopathy, 605899
ANK3	162.1	99%	99%	?Mental retardation, autosomal recessive, 37
ANKH	104.1	100%	100%	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKRD11	85.4	96%	91%	KBG syndrome, 148050

ANO10	108.7	96%	95%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	127.4	97%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S2	53.2	66%	56%	Mental retardation, X-linked syndromic, Fried type, 300630
AP3B1	93.9	99%	92%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	143	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	103.9	99%	96%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	93.2	100%	93%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	72.8	73%	68%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	122.4	92%	86%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	154.8	98%	97%	Periventricular heterotopia with microcephaly, 608097
ARG1	149.5	100%	100%	Argininemia, 207800
ARHGEF6	99.8	96%	89%	Mental retardation, X-linked 46, 300436
ARHGEF9	64.2	100%	99%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	135.8	91%	87%	Mental retardation, autosomal dominant 14, 614607
ARID1B	131.2	92%	84%	Mental retardation, autosomal dominant 12, 614562
ARID2	216.5	97%	95%	No OMIM phenotype Intellectual disability (Shang (2015) Neurogenetics epub) Schizophrenia (Fromer (2014) Nature 506,179)
ARL13B	78.8	99%	81%	Joubert syndrome 8, 612291
ARL6	94.9	99%	96%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARSE	58.9	96%	86%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	18.6	62%	41%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia,300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510
ASL	88.5	100%	96%	Argininosuccinic aciduria, 207900
ASNS	80.6	96%	91%	Asparagine synthetase deficiency, 615574
ASPA	148.4	100%	96%	Canavan disease, 271900
ASPM	95.3	96%	91%	Microcephaly 5, primary, autosomal recessive, 608716

ASXL1	145	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	150.2	98%	97%	Bainbridge-Ropers syndrome, 615485
ATIC	114.3	100%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A2	176.3	100%	100%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481
ATP2A2	166.4	100%	99%	Darier disease, 124200 Acrokeratosis verruciformis, 101900
ATP6AP2	27.9	64%	41%	Mental retardation, X-linked, with epilepsy, 300423
ATP6V0A2	141.9	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	88.3	99%	93%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	131.6	100%	100%	?Cerebellar ataxia, mental retardation and dysequilibrium syndrome 4, 615268
ATR	137.4	97%	95%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	54.3	96%	88%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	64.3	93%	89%	3-methylglutaconic aciduria, type I, 250950
AUTS2	92.9	96%	94%	Mental Retardation, autosomal dominant 26, 615834
B3GALTL	100.3	95%	94%	Peters-plus syndrome, 261540
B4GALT1	110.9	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	96.5	95%	94%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
BBS1	133.8	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	160.2	100%	99%	Bardet-Biedl syndrome 10, 209900
BBS12	196.3	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	173	100%	98%	Bardet-Biedl syndrome 2, 209900
BBS4	136.1	99%	95%	Bardet-Biedl syndrome 4, 209900
BBS5	103.3	97%	91%	Bardet-Biedl syndrome 5, 209900
BBS7	121.3	96%	90%	Bardet-Biedl syndrome 7, 209900
BBS9	105.1	94%	93%	Bardet-Biedl syndrome 9, 209900

BCKDHA	147.8	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	120.7	85%	81%	Maple syrup urine disease, type Ib, 248600
BCL11A	116.6	97%	97%	No OMIM phenotype Autism spectrum disorder & developmental delay (Basak (2015) J Clin Invest 125, 2363) Childhood apraxia of speech,disarthria,oral/motor dyspraxia,hypotonia and intellectual delay (Peter (2014) Am J Med Genet A 164,2091) Developmental
BCOR	71	98%	95%	Microphthalmia, syndromic 2, 300166
BCS1L	147	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BLM	115.5	98%	92%	Bloom syndrome, 210900
BRAF	68.9	89%	83%	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
BRWD3	65.9	96%	90%	Mental retardation, X-linked 93, 300659
BSCL2	107.4	100%	98%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
BTD	122.5	100%	99%	Biotinidase deficiency, 253260
BUB1B	143.9	98%	97%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf57	124.3	100%	100%	Temtamy syndrome, 218340
C12orf65	74.3	99%	97%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C2CD3				?Orofaciodigital syndrome XIV,615948
C5orf42	119.9	95%	91%	Joubert syndrome 17, 614615
CA2	130.8	90%	85%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

CA8	98.2	87%	87%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNG2	118.1	100%	100%	Mental retardation, autosomal dominant 10, 614256
CAMTA1	155.6	99%	97%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CASK	63.1	99%	90%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CBL	118.2	98%	96%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	106.9	99%	91%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	103.6	100%	98%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	121.8	99%	94%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCBE1	69.9	100%	95%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC78	92.3	100%	98%	Myopathy, centronuclear, 4, 614807
CDH15	111.1	100%	95%	Mental retardation, autosomal dominant 3, 612580
CDK5RAP2	127.9	99%	96%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	83	95%	91%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CDON	132.5	100%	98%	Holoprosencephaly 11, 614226
CENPJ	144.5	99%	96%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	73.6	94%	85%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	181.5	97%	94%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	69.5	89%	76%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	83.4	97%	91%	Joubert syndrome 15, 614464
CHAMP1	143.4	100%	100%	Mental retardation,autosomal dominant 40,616579
CHD2	124.9	99%	98%	Epileptic encephalopathy, childhood-onset, 615369

CHD7	136.1	99%	96%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	152	100%	99%	{Autism, susceptibility to, 18}, 615032
CHKB	82.4	98%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CLCNKB	87.7	95%	89%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLIC2	50.1	90%	81%	Mental retardation, X-linked, syndromic 32, 300886
CLN3	107.2	99%	97%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	144.8	97%	91%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	119.2	100%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	206.6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNKSR2	62.9	95%	79%	No OMIM phenotype Intellectual disability,X-linked non syndromic (Vaags (2014) Ann Neurol 76,758)
CNTNAP2	130	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COG1	114.1	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG6	78.4	89%	79%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	120.1	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	92.3	100%	90%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	83	96%	89%	?Retinal arteries,tortuosity of,180000 Angiopathy,hereditary,with nephropathy,aneurysms and muscle cramps,611773 Brain small vessel disease with or without ocular anomalies,607595 Porencephaly 1,175780 {Hemorrhage,intracerebral,susceptibility to},614519
COL4A2	83.6	98%	92%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	124.9	99%	95%	Mental retardation, autosomal dominant 34, 616351
COLEC11	179.7	100%	100%	3MC syndrome 2, 265050
COQ2	71.1	97%	91%	Coenzyme Q10 deficiency, primary, 1, 607426

				{Multiple system atrophy, susceptibility to}, 146500
COQ4	80.5	84%	81%	Coenzyme Q10 deficiency,primary,7,616276
COX10	202	100%	98%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	94.5	100%	99%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
CPS1	148.8	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CRADD	108.1	100%	93%	Mental retardation, autosomal recessive 34, 614499
CRBN	135.5	100%	97%	Mental retardation, autosomal recessive 2, 607417
CREBBP	119.9	99%	96%	Rubinstein-Taybi syndrome, 180849
CSNK2A1	128.4	91%	84%	No OMIM phenotype Glaucoma, primary congenital (Lee (2011) Mol Vis 17,3583)
CTCF	143.8	98%	95%	Mental retardation, autosomal dominant 21, 615502
CTDP1	93.2	90%	85%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNB1	163.2	100%	100%	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNND1	153.1	100%	99%	No OMIM phenotype Autism? (O'Roak (2012) Nature 485,246)
CTSA	126.3	99%	96%	Galactosialidosis, 256540
CTSD	152.8	98%	94%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	137.2	98%	94%	No OMIM phenotype Autism? (Iossifov (2012) Neuron 74,285)
CUBN	124.5	99%	97%	Megaloblastic anemia-1, Finnish type, 261100
CUL4B	47.4	93%	84%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CYB5R3	152.2	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
D2HGDH	116.5	97%	93%	D-2-hydroxyglutaric aciduria, 600721
DARS2	117.7	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	106.5	99%	87%	Maple syrup urine disease, type II, 248600

DCAF17	92.2	99%	92%	Woodhouse-Sakati syndrome, 241080
DCPS				Al-Raqad syndrome,616459
DCX	76.6	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDHD2	160.8	99%	94%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	83.4	79%	70%	Warsaw breakage syndrome, 613398
DDX3X	62.4	94%	86%	Mental retardation,X-linked 102,300958
DEAF1	121.3	91%	83%	Mental retardation, autosomal dominant 24, 615828
DHCR24	159.3	100%	100%	Desmosterolosis, 602398
DHCR7	149.9	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHFR	65.1	94%	89%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	139.3	98%	97%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIAPH1	114.3	99%	97%	Deafness,autosomal dominant 1,124900 Seizures,cortical blindness,microcephaly syndrome,616632
DIP2B	165.3	98%	96%	Mental retardation, FRA12A type, 136630
DKC1	77.4	99%	92%	Dyskeratosis congenita, X-linked, 305000
DLD	129.1	98%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	60.4	99%	86%	Mental retardation, X-linked 90, 300850
DLG4	163	100%	98%	no OMIM phenotype Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394)
DMD	83.9	99%	94%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DMPK	98.4	100%	93%	Myotonic dystrophy 1, 160900
DNAJC19	96.9	99%	88%	3-methylglutaconic aciduria, type V, 610198
DNM1	142.4	91%	90%	Epileptic encephalopathy,early infantile,31,616346
DNMT3A	98	96%	92%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	122.9	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK8	129.4	100%	99%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DPAGT1	112.3	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	112.1	89%	84%	Congenital disorder of glycosylation, type Ie, 608799

DPP6	132.1	98%	94%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}
DPYD	166.8	97%	93%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DST	159.5	99%	97%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DYM	105	97%	95%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326 Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DYNC1H1	166.1	99%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	153.5	100%	99%	Mental retardation, autosomal dominant 7, 614104
EBP	71	97%	89%	Chondrodysplasia punctata, X-linked dominant, 302960
EDC3	139.9	100%	99%	?Mental retardation, autosomal recessive 50,616460
EEF1A2	176.2	100%	100%	Epileptic encephalopathy, early infantile,33,616409 Mental retardation,autosomal dominant 38,616393
EFTUD2	114.2	99%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	130.5	99%	97%	Kleefstra syndrome, 610253
EIF2AK3	152.8	97%	89%	Wolcott-Rallison syndrome, 226980
EIF4G1	119.8	100%	99%	Parkinsons disease 18, 614251
ELOVL4	77.5	100%	98%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMX2	105	100%	99%	Schizencephaly, 269160
EP300	175.7	100%	99%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPB41L1	121.4	100%	100%	Mental retardation, autosomal dominant 11, 614257
ERCC2	120	100%	99%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	94.3	100%	99%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC5	133.9	100%	96%	Xeroderma pigmentosum, group G, 278780

				Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	165.9	100%	100%	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	86.8	90%	83%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	151.4	100%	99%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	100	91%	86%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	106.2	100%	100%	Glutaric acidemia 2B, 231680
ETHE1	64.8	99%	88%	Ethylmalonic encephalopathy, 602473
EXOSC3	62.6	97%	88%	Pontocerebellar hypoplasia, type 1B, 614678
EZH2	131.8	99%	96%	Weaver syndrome, 277590
FAM126A	119.4	99%	94%	Leukodystrophy, hypomyelinating, 5, 610532
FBN1	161.3	99%	98%	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
FBXO31	89.3	93%	89%	Mental retardation, autosomal recessive 45, 615979
FGD1	53.8	89%	77%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400

FGFR1	135.5	100%	95%	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	133.9	96%	95%	Antley-Bixler syndrome wo 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,mental retardation,609579
FGFR3	91.3	100%	98%	Achondrodysplasia,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	160.3	90%	87%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIGN	136.4	100%	100%	No OMIM phenotype
FKRP	70.4	100%	96%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital w/wo mental retardation), type B, 5, 606612
FKTN	137.2	97%	89%	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	85.1	99%	97%	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	121.4	99%	97%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FMN2	87.1	84%	77%	Mental retardation, autosomal recessive 47, 616193
FMR1	51.7	86%	75%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FOXG1	88.7	85%	81%	Rett syndrome, congenital variant, 613454
FOXP1	123.6	100%	99%	Mental retardation with language impairment and autistic features, 613670
FOXP2	156.4	98%	94%	Speech-language disorder-1, 602081
FRAS1	142.2	100%	99%	Fraser syndrome, 219000
FTO	154.4	99%	98%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	78.7	96%	92%	Mental retardation, X-linked 9, 309549
FUCA1	120.8	99%	98%	Fucosidosis, 230000
GABRA1	176.6	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4} {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136

GAD1	116.9	99%	93%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALE	141.6	100%	100%	Galactose epimerase deficiency, 230350
GALT	141.5	100%	99%	Galactosemia, 230400
GAMT	94.4	98%	91%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	122.4	100%	99%	Mental retardation, autosomal dominant 18, 615074
GATM	152.2	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	68.7	87%	79%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	38.9	93%	61%	Glycine encephalopathy, 605899
GDI1	103.7	100%	100%	Mental retardation, X-linked 41, 300849
GFAP	87.2	100%	97%	Alexander disease, 203450
GJB1	114.6	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	34.3	81%	62%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GK	29.1	66%	50%	Glycerol kinase deficiency, 307030
GLB1	73.9	98%	90%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	77.2	87%	79%	Glycine encephalopathy, 605899
GLI2	110.3	97%	93%	Holoprosencephaly-9, 610829
GLI3	138.5	100%	100%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200 {Hypothalamic hamartomas, somatic}, 241800
GM2A	128.7	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPA	124.4	100%	100%	Alacrima, achalasia and mental retardation syndrome, 615510
GMPPB	206.3	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	173.4	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473

GNAS	110.5	95%	94%	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 Pseudopseudohypoparathyroidysm,612463
GNPAT	138.4	99%	96%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNS	104.7	92%	88%	Mucopolysaccharidosis type IIID, 252940
GPC3	72	93%	85%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	172.9	96%	96%	Molybdenum cofactor deficiency, type C, 252150
GPR56	140.4	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854
GPT2	134.6	100%	98%	?Mental retardation, autosomal recessive 49, 616281
GRIA3	65.5	96%	85%	Mental retardation, X-linked 94, 300699
GRID2	177	100%	98%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	130.6	95%	89%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	132.6	100%	98%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	157.9	100%	100%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	184.8	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRIN3B	88.1	83%	76%	No OMIM phenotype Schizophrenia, increased risk,association with (Matsuno (2015) PLoS One 10,e0116319)
GRM1	169.3	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSE1	77.6	99%	97%	No OMIM phenotype Autism (Sanders (2012) Nature 485,237)
GSS	92.9	100%	100%	Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	160.4	100%	99%	Trichothiodystrophy, complementation group A, 601675
GUSB	105.3	89%	85%	Mucopolysaccharidosis VII, 253220
HACE1				Spastic paraplegia and psychomotor retardation with or without seizures,616756
HAX1	123.9	100%	99%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	63.8	99%	92%	Microphthalmia, syndromic 7, 309801
HCFC1	64.6	97%	91%	Mental retardation, X-linked 3, 309541
HCN1	117.4	100%	100%	Epileptic encephalopathy, early infantile, 24, 615871

HDAC4	94.5	100%	99%	Brachydactyly-mental retardation syndrome, 600430
HDAC6	65.9	96%	87%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	90.5	100%	100%	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882
HECTD1	169.3	98%	96%	No OMIM phenotype
HERC1	173.3	100%	99%	No OMIM phenotype Overgrowth,intellectual disability and facial dysmorfism (Ortega-Recalde (2015) Clin Genet 88,e1)
HERC2	98.8	77%	72%	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	65.2	99%	85%	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	125.4	100%	99%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	134.4	97%	91%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HIVEP2	181.9	100%	100%	No OMIM phenotype Intellectual disability,nonsyndromic (Rauch (2012) Lancet epub)
HLCS	160.7	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNMT				Mental retardation,autosomal recessive 51,616739 {Asthma,susceptibility to},600807
HOXA1	125.4	100%	100%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HPD	131.5	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	45.8	95%	84%	Lesch-Nyhan syndrome, 300322
HRAS	165.7	100%	98%	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome,somatic mosaic,163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous, somatic}, 162900 {Spitz nevus or nevus spilus,somatic},137550 {Thyroid carcinoma, follicular, somatic}, 188470

HSD17B10	71.2	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSPD1	84.8	97%	89%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HUWE1	66	98%	94%	Mental retardation, X-linked syndromic, Turner type, 300706
IDS	62.5	98%	98%	Mucopolysaccharidosis II, 309900
IDUA	94.1	89%	85%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IER3IP1	55.9	98%	85%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFT172	108.6	99%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IGBP1	75.4	96%	90%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	124.4	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	25.6	68%	55%	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33,300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, type II, 308300 Invasive pneumococcal disease,recurrent isolated,2,300640
IL1RAPL1	77.5	100%	92%	Mental retardation, X-linked 21/34, 300143
INPP5E	83.9	96%	88%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
IQSEC2	37.5	87%	76%	Mental retardation, X-linked 1, 309530
ISPD	107.2	96%	82%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITPR1	157.5	100%	99%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
JAG1	139.8	100%	97%	Alagille syndrome, 118450 Deafness, congenital heart defects and posterior embryotoxon Tetralogy of Fallot, 187500
JAM3	136.4	100%	99%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANK1	155.8	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	70.8	96%	89%	Koolen-De Vries syndrome, 610443
KAT6A	161.2	100%	99%	Mental retardation, autosomal dominant 32, 616268

KAT6B	167.3	98%	97%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNC3	140.8	64%	53%	Spinocerebellar ataxia 13,605259
KCNH1	185.4	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	199.6	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	257	100%	100%	Diabetes mellitus,permanent neonatal,with neurologic features,606176 Diabetes mellitus,transient neonatal,3,610582 Hyperinsulinemic hypoglycemia,familial,2,601820 Maturity-onset diabetes of the young,type 13,616329 {Diabetes mellitus,type 2,susceptibility to},125853
KCNK9	164.3	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	80.5	98%	92%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KCNQ5	139.9	98%	97%	No OMIM phenotype Schizophrenia (Fromer (2014) Nature 506,179)
KCNT1	106.6	94%	91%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCTD7	113.5	95%	93%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A				Cleft palate,psychomotor retardation,and distinctive facial features,616728
KDM5C	68.6	95%	90%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	74.7	88%	75%	Kabuki syndrome 2, 300867
KIAA0226	101.9	97%	97%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA1033	81.5	94%	87%	?Mental retardation, autosomal recessive 43, 615817
KIAA1109	143.5	97%	95%	no OMIM phenotype Dandy-Walker malformation,hydrocephalus,flexed deformity,club feet,micrognathia and pleural effusion (Alazami (2015) Cell Rep 10,148) Schizophrenia (Gulsuner (2013) Cell 154,518)
KIAA1279	165.3	100%	100%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	99.9	100%	98%	Mental retardation, X-linked 98, 300912
KIF11	76.6	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950

KIF1A	107.9	98%	94%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF4A	58.3	91%	82%	?Mental retardation,X-linked 100,300923
KIF5C	104.9	98%	97%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	70	91%	84%	?Al-Gazali-Bakalinova syndrome,607131 ?Hydrolethalus syndrome 2,614120 Acrocallosal syndrome,200990 Joubert syndrome 12,200990
KIRREL3	132.8	100%	98%	Mental retardation, autosomal dominant 4, 612581
KMT2A	142.8	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	133.1	99%	99%	Kabuki syndrome 1, 147920
KPTN	88.6	100%	99%	Mental retardation, autosomal recessive 41, 615637
KRAS	57.8	100%	99%	Bladder cancer,somatic,109800 Breast cancer,somatic,114480 Cardiofaciocutaneous syndrome 2,615278 Gastric cancer,somatic,137215 Leukemia,acute myeloid,601626 Lung cancer,somatic,211980 Noonan syndrome 3,609942 Pancreatic carcinoma, somatic,260350 RAS-associated autoimmune leukoproliferative disorder,614470 Schimmelpenning-Feuerstein-Mins syndrome,somatic mosaic,163200
KRBOX4	117	100%	100%	No OMIM phenotype nonsyndromic X-linked mental retardation (Lugtenberg et al. 2006)
L1CAM	87.3	98%	95%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000
L2HGDH	120.8	97%	95%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	134.2	99%	98%	Poretti-Boltshauser syndrome, 615960
LAMA2	144.2	99%	98%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855

LAMC3	112.2	98%	92%	Cortical malformations, occipital, 614115
LAMP2	76.4	92%	91%	Danon disease, 300257
LARGE	122.9	100%	97%	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	48.3	71%	58%	Alazami syndrome, 615071
LIG4	163.2	100%	98%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LINS	131.6	99%	96%	Mental retardation, autosomal recessive 27, 614340
LRP2	176.4	100%	99%	Donnai-Barrow syndrome, 222448
LRPPRC	130	98%	94%	Leigh syndrome, French-Canadian type, 220111
MAGEL2	114.5	100%	100%	Prader-Willi-like syndrome, 615547
MAGT1	71	97%	84%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAN1B1	125.7	100%	100%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	103.9	97%	91%	Mannosidosis, alpha-, types I and II, 248500
MANBA	120.6	99%	93%	Mannosidosis, beta, 248510
MAOA	81.3	98%	96%	Brunner syndrome, 300615
MAP2K1	89.5	100%	98%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	103.9	94%	90%	Cardiofaciocutaneous syndrome 4, 615280
MAT1A	176.9	97%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	179.2	100%	100%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	84.9	99%	91%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MCCC1	147.3	100%	100%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	120.8	99%	96%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	130.6	97%	95%	Mucolipidosis IV, 252650
MCPH1	133.9	99%	94%	Microcephaly 1, primary, autosomal recessive, 251200

MECP2	48.4	97%	75%	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	64.3	95%	89%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	131.9	100%	98%	Mental retardation and distinctive facial features with or without cardiac defects,616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	129.9	99%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	131	99%	97%	Mental retardation, autosomal recessive 18, 614249
MEF2C	112.8	99%	88%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443
METTL23	141.3	100%	100%	Mental retardation, autosomal recessive 44, 615942
MGAT2	137.6	100%	99%	Congenital disorder of glycosylation, type IIa, 212066
MID1	111.1	100%	99%	Opitz GBBB syndrome, type I, 300000
MID2	94.3	98%	94%	?Mental retardation,X-linked 101,300928
MKKS	222.1	89%	89%	Bardet-Biedl syndrome 6, 209900 McKusick-Kaufman syndrome, 236700
MLYCD	71	93%	90%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	177.2	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMACHC	173.1	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	79.1	83%	55%	Homocystinuria, cbID type, 277410
MOCS1	73	96%	91%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	145.7	99%	99%	Molybdenum cofactor deficiency, type B, 252150
MOGS	99.7	99%	98%	Congenital disorder of gycosylation, type 2b, 606056
MPDU1	113.6	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPDZ	156.9	98%	96%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPLKIP	76.3	88%	52%	Trichothiodystrophy, nonphotosensitive 1, 234050
MRPS22	129.9	93%	90%	Combined oxidative phosphorylation deficiency 5, 611719

MTHFR	134.9	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTOR				Smith-Kingsmore syndrome,616638
MTR	132.8	99%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	118.7	100%	97%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MUT	113.3	97%	91%	Methylmalonic aciduria, mut(0) type, 251000
MVK	126.3	100%	100%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYCN	68.2	90%	78%	Feingold syndrome, 164280
MYH9	123.8	99%	95%	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	122.5	98%	95%	Griscelli syndrome, type 1, 214450
MYT1L	171.5	99%	99%	Mental retardation,autosomal dominant 39,616521
NAA10	58	100%	91%	N-terminal acetyltransferase deficiency, 300855
NAGA	147.9	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	99.9	91%	88%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	136.2	99%	96%	?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419
NBN	74.1	99%	96%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NDE1	77	100%	100%	Lissencephaly 4 (with microcephaly), 614019
NDP	80.3	100%	100%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390

NDST1	182	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	138.7	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFA11	74.8	98%	91%	Mitochondrial complex I deficiency, 252010
NDUFA12	138	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFS1	134.3	97%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	102.5	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	121.8	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	155.3	100%	99%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS7	106.9	100%	99%	Leigh syndrome, 256000
NDUFS8	125.5	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	132.1	100%	97%	Mitochondrial complex I deficiency, 252010
NEDD4L	137.3	100%	97%	No OMIM phenotype 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) Essential hypertension,association with (Russo (2005) Hypertension 46,488)
NEU1	16.3	66%	31%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NF1	122.7	91%	86%	Neurofibromatosis, type 1, 162200
NFATC1	97.7	99%	94%	No OMIM phenotype Tricuspid atresia (Abdul-Sater (2012) PLoS One 7,e49532) Congenital heart disease (Glnessner (2014) Circ Res 115,884)
NFIA	121	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)
NFIX	139.2	98%	94%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NHS	83.7	93%	88%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIPBL	111.4	96%	93%	Cornelia de Lange syndrome 1, 122470

NKX2-1	47.7	98%	80%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer,nonmedullary,1},188550
NLGN3	84.1	100%	95%	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	113.8	100%	95%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility,X-linked 2},300495
NLRP3	130.4	100%	100%	CINCA syndrome, 607115 Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900
NPHP1	122	99%	94%	Joubert syndrome 4,609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1,266900
NR2F1	182.2	99%	92%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRAS	179.8	100%	100%	?RAS-associated autoimmune lymphoproliferative syndrome type IV,somatic,611470 Colorectal cancer,somatic,114500 Epidermal nevus,somatic,162900 Melanocytic nevus syndrome,congenital,somatic,137550 Neurocutaneous melanosis,somatic,249400 Noonan syndrome 6,613224 Schimmelpenning-Feuerstein-Mims syndrome,somatic mosaic,163200 Thyroid carcinoma,follicular,somatic,188470
NRXN1	160	99%	96%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	142.2	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	119.2	99%	93%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	108.9	95%	90%	Mental retardation, autosomal recessive 5, 611091
NTRK1	115.4	97%	93%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
OCLN	164.9	100%	99%	Band-like calcification with simplified gyration and polymicrogyria, 251290

OCRL	87	97%	88%	Lowe syndrome, 309000 Dent disease 2, 300555
ODC1	138.4	100%	99%	{Colonic adenoma recurrence,reduced risk of},114500
OFD1	29.6	74%	53%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome type 2,300209
OPHN1	63.9	98%	94%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	104.1	100%	97%	Meier-Gorlin syndrome 1, 224690
OTC	87	100%	99%	CGD Ornithine transcarbamylase deficiency, 311250
PACS1	108.7	95%	94%	Mental retardation, autosomal dominant 17, 615009
PAFAH1B1	97.6	89%	80%	Lissencephaly, 607432 Subcortical laminar heterotopia, 607432
PAH	170.6	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	53	96%	80%	Mental retardation, X-linked 30/47, 300558
PANK2	142.9	95%	86%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX1	105.6	83%	77%	?Orofaciocervical syndrome 2, 615560
PAX6	129.3	100%	100%	?Morning glory disc anomaly, 120430 Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve,120430 Coloboma,ocular,120200 Foveal hyperplasia, 136520 Keratitis, 148190 Optic nerve hypoplasia,165550 Peters anomaly, 604229 Optic nerve hypoplasia, 165550
PAX8	74.5	100%	99%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PC	134.1	98%	94%	Pyruvate carboxylase deficiency, 266150

PCDH19	133.3	100%	95%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	99.9	100%	98%	no OMIM phenotype Developmental disorder (Fitzgerald (2015) Nature 519,223)
PCNT	108.1	97%	93%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	112.7	97%	95%	Acrocydostosis 2 with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDHA1	72.7	94%	82%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDSS1	118.2	92%	83%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	109	97%	92%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	99.8	100%	97%	Prolidase deficiency, 170100
PEX1	106.9	97%	97%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	100.3	97%	87%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	107.2	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	138.3	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	194.6	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	114	93%	91%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	100	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	73.3	100%	98%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	97	100%	99%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	100.1	98%	94%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	75.8	86%	82%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	121.4	91%	82%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGAP1	97.6	95%	92%	Mental retardation, autosomal recessive 42, 615802

PGAP2	156.2	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	113.5	97%	94%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	35.1	80%	65%	Phosphoglycerate kinase 1 deficiency, 300653
PHF6	46.3	87%	81%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	65.5	98%	92%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	119.4	100%	99%	Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	126.2	96%	92%	No OMIM phenotype Intellectual disability (de Ligt (2012) N Engl J Med 367,1921) Glaucoma,primary congenital (Lee (2011) Mol Vis 17,3583)
PIGL	111.9	100%	99%	CHIME syndrome, 280000
PIGN	116.9	96%	90%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	113.4	100%	98%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	145.2	99%	98%	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	149.2	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3R2	74.1	87%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PLA2G6	108.7	100%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLCB1	153.4	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	91.3	100%	99%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLXND1	113.1	97%	92%	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6) Truncus arteriorus (Ta-Shma (2013) Am J Med Genet A 161,3115) Diabetic nephropathy,association with (McKnight (2009) Hugo J 3,77)
PMM2	163.1	100%	98%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	78.6	100%	95%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	125.9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POC1A	132.6	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POGZ	159.4	99%	98%	White-Sutton syndrome,616364
POLG	107.7	100%	98%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome, 607459

				Progressive external ophthalmoplegia, autosomal dominant, 157640
POLR3A	140.2	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	143.9	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	107	100%	95%	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
POMK				?Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,12,616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,12,615249
POMT1	152.8	100%	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	101.6	97%	95%	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	79.8	100%	96%	Focal dermal hypoplasia, 305600
POU1F1	118.2	99%	92%	Pituitary hormone deficiency, combined, 1, 613038
PPOX	92.1	100%	97%	Porphyria variegata, 176200
PPP2R1A	124.1	93%	93%	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	131.9	100%	100%	Mental retardation, autosomal dominant 35, 616355
PPT1	150.6	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	97	97%	94%	Renpenning syndrome, 309500
PRODH	68.5	84%	77%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	113.7	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	154.4	98%	96%	Mental retardation, autosomal recessive 1, 249500

PSAP	111.2	100%	97%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSEN1	140.9	98%	94%	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
PTCH1	116.3	98%	96%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTCHD1	106.5	100%	97%	{Autism,susceptibility to,X-linked 4},300830
PTDSS1	130.4	100%	99%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	142.2	100%	100%	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	93	96%	89%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PUF60	140.1	99%	97%	Verheij syndrome, 615583
PURA	88.9	89%	85%	Mental retardation, autosomal dominant 31, 616158
PUS1	130	99%	97%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	80.4	100%	91%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB18	88.4	97%	78%	Warburg micro syndrome 3, 614222

RAB27A	177.8	100%	100%	Griscelli syndrome, type 2, 607624
RAB39B	89.2	99%	95%	Mental retardation, X-linked 72, 300271
RAB3GAP1	127.3	99%	99%	Warburg micro syndrome 1, 600118
RAB3GAP2	94	98%	93%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	100	100%	100%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519
RAC1	116.7	99%	90%	No OMIM phenotype
RAD21	89.1	99%	96%	Cornelia de Lange syndrome 4, 614701
RAF1	118.6	100%	99%	Cardiomyopathy,dilated,1NN,615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAI1	113.3	100%	99%	Immunodeficiency 9, 612782 Smith-Magenis syndrome, 182290
RARS2	111.7	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RBM10	67.6	99%	94%	TARP syndrome, 311900
RBM28	128.8	100%	100%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RELN	158.4	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320
REV3L	142.4	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	93.3	100%	91%	Congenital disorder of glycosylation, type In, 612015
RHEB	32.2	88%	64%	No OMIM phenotype
RIT1	154.3	100%	100%	Noonan syndrome 8, 615355
RMND1	131.5	99%	90%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP				Anauxetic dysplasia,607095 Cartilage-hair hypoplasia,250250 Metaphyseal dysplasia without hypotrichosis,250460
RNASEH2A	124.1	100%	95%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	106.7	88%	76%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	175.9	100%	99%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	74.6	83%	80%	Leukoencephalopathy, cystic, without megalencephaly, 612951
ROGDI	110.8	96%	94%	Kohlschutter-Tonz syndrome, 226750

RPGRIP1L	133.2	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	50.2	99%	78%	{Autism, susceptibility to, X-linked 5}, 300847
RPS6KA3	55.2	88%	80%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RTEL1	100	98%	92%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
SALL1	115.7	100%	98%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SATB2	98	98%	91%	Cleft palate and mental retardation, 119540
SC5D	199.5	100%	100%	Lathosterolosis, 607330
SCN1A	151.4	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	153.2	99%	97%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN8A	192.7	99%	97%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCO2	94.1	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	96.8	81%	73%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SERAC1	112.7	99%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	115.9	97%	96%	Schinzel-Giedion midface retraction syndrome, 269150
SETD2	143.8	100%	99%	Luscan-Lumish syndrome,616831
SETD5	176.3	100%	99%	Mental retardation,autosomal dominant 24,615761
SF1	72.9	86%	78%	No OMIM phenotype
SGSH	120.8	94%	92%	Mucopolysaccharidisis type 3A (Sanfilippo A), 252900
SHANK2	120.3	100%	99%	{Autism susceptibility 17}, 613436
SHANK3	65.5	79%	70%	Phelan-McDermid syndrome, 606232

				{Schizophrenia 15}, 613950
SHH	84.8	95%	91%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	124.9	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	58.9	99%	90%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIL1	149.3	97%	96%	Marinesco-Sjogren syndrome, 248800
SIN3A	148.4	99%	98%	No OMIM phenotype Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
SIX3	122.8	97%	89%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SKI	68.1	96%	95%	Shprintzen-Goldberg syndrome, 182212
SLC12A6	144.7	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	34.9	90%	77%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	104.2	99%	92%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC19A3	168.9	100%	100%	Thiamine metabolism dysfunction syndrome 2(biotin- or thiamine-responsive encephlopathy type 2),607483
SLC1A1	188.8	100%	98%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A4	147.2	100%	97%	Spastic tetraplegia,thin corpus callosum,and progressive microcephaly,616657
SLC25A15	212.9	98%	97%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	91.8	100%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	161.8	100%	98%	Dystonia 9,601042 GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects,608885 {Epilepsy, idiopathic generalized,susceptibility,12},614847
SLC33A1	133.8	98%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A2	66.2	97%	93%	Congenital disorder of glycosylation, type 2m, 300896
SLC35C1	198.2	99%	96%	Congenital disorder of glycosylation, type IIc, 266265
SLC39A12	103.3	99%	93%	No OMIM phenotype

SLC4A4	132.4	100%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A17	187.6	100%	100%	Mental retardation, autosomal recessive 48, 616269
SLC6A3	122.6	100%	99%	Parkinsonism -dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	32.5	79%	66%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	106.3	100%	100%	Lysinuric protein intolerance, 222700
SLC9A6	73.9	93%	83%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	114.6	99%	99%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer,somatic,260350 Polyposis, juvenile intestinal, 174900
SMARCA2	109.5	96%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	131.9	99%	94%	Mental retardation, autosomal dominant 16, 614609 Rhabdoid tumor predisposition syndrome 2, 613325
SMARCB1	202.6	100%	100%	Coffin-Siris syndrome 3,614608 Rhabdoid tumors,somatic,609322 {Rhabdoid predisposition syndrome 1},609322 {Schwannomatosis-1,susceptibility to},162091
SMARCC2	111.2	96%	95%	No OMIM phenotype Ivemark syndrome (Carss (2014) Hum Mol Genet 23,3269) Autism (Neale (2012) Nature 485,242)
SMARCE1	69.9	94%	85%	{Meningioma, familial, susceptibility to}, 607174
SMC1A	67.1	99%	95%	Cornelia de Lange syndrome 2, 300590
SMC3	77.3	94%	77%	Cornelia de Lange syndrome 3, 610759
SMOC1	107.8	100%	96%	Microphthalmia with limb anomalies, 206920
SMPD1	110.9	100%	97%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	40.3	85%	61%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP29	134.3	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	125.3	97%	95%	Psychomotor retardation, epilepsy and craniofacial dysmorphism, 614501
SNX14	69.9	93%	77%	Spinocerebellar ataxia,autosomal recessive 20,616354
SOBP	93	90%	77%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671

SON	146.1	97%	92%	No OMIM phenotype Schizophrenia (Fromer (2014) Nature 506,179) Developmental delay,seizure disorder,macrocephaly and white matter abnormalities (Zhu (2015) Genet Med)
SOS1	87.5	93%	88%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SOX10	65.7	92%	88%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	109.3	100%	94%	Mental retardation, autosomal dominant, 27, 615866
SOX2	80.8	99%	96%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	17.5	81%	38%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	115.1	99%	97%	Lamb-Shaffer syndrome, 616803
SPG11	130.1	98%	95%	Spastic paraplegia 11, autosomal recessive, 604360
SPRED1	165.6	96%	96%	Legius syndrome, 611431
SPTAN1	116.4	99%	97%	Epileptic encephalopathy, early infantile, 5
SRCAP	131.2	99%	98%	Floating-Harbor syndrome, 136140
SRD5A3	140.5	99%	96%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	51.4	98%	95%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	163.4	100%	100%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	118.3	94%	93%	Amish infantile epilepsy syndrome, 609056
STAG1	110.7	99%	97%	No OMIM phenotype Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)
STIL	135.4	100%	99%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	105.6	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STT3A	161.5	100%	100%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	112.2	97%	93%	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	160.2	100%	99%	Generalized epilepsy with febrile seizures plus,type 9,616172
STXBP1	124.2	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164

SUCLA2	58	94%	76%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUOX	189.8	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	87.3	88%	88%	Leigh syndrome, due to COX deficiency, 256000
SYN1	44.3	70%	56%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	46.7	89%	76%	No OMIM phenotype Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)
SYNE1	137.1	99%	98%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNGAP1	60.9	92%	79%	Mental retardation, autosomal dominant 5, 612621
SYP	40	93%	74%	Mental retardation, X-linked 96, 300802
SYT14	174.6	93%	83%	Spinocerebellar ataxia, autosomal recessive 11, 614229
TAF2	121	99%	95%	Mental retardation, autosomal recessive 40, 615599
TAT	117.2	100%	100%	Tyrosinemia, type II, 276600
TBC1D24	149.2	100%	100%	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	99.3	95%	91%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	147.1	99%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome-1, 244460
TBR1	98.4	99%	94%	No OMIM phenotype Intellectual disability (Hamdan (2014) PLoS Genet 10) Autism (O'Roak (2012) Science 338,1619) Ventriculomegaly (Traylor (2012) Mol Syndromol 3,102)
TCF20	136.7	100%	100%	No OMIM phenotype Autism spectrum disorder (Babbs (2014) J Med Genet 51,737)
TCF4	137.6	100%	100%	Pitt-Hopkins syndrome, 610954
TCF7L2	135.6	96%	90%	{Diabetes mellitus,type 2,susceptibility to},125853
TECR	80.7	98%	91%	Mental retardation, autosomal recessive 14, 614020
TFAP2A	109.5	100%	98%	Branchiooculofacial syndrome, 113620
TGFBR1	196.2	93%	93%	Loeys-Dietz syndrome, type 1, 609192 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800

TGFBR2	184.5	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 2, 610168
TGIF1	126.4	100%	97%	Holoprosencephaly-4, 142946
TH	66.4	95%	87%	Segawa syndrome,recessive,605407
THOC6	227.3	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	165.9	100%	98%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	17.5	64%	30%	Mohr-Tranebjaerg syndrome, 304700
TLK2	86.4	94%	87%	No OMIM phenotype Schizophrenia (Gulsuner (2013) Cell 154,518)
TMCO1	81	100%	98%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM165	107.3	99%	96%	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	86.1	99%	95%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	96.1	99%	97%	Joubert syndrome 14, 614424
TMEM67	70.3	91%	84%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TMLHE	63.4	100%	86%	Epsilon-trimethyllysine hydrolylase deficiency, 300872
TPP1	124	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TRAPPC11	121	96%	95%	Muscular dystrophy, limb-girdle, type 2S
TRAPPC9	124	100%	99%	Mental retardation, autosomal recessive 13, 613192
TREX1	214.3	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	134.2	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110
TRIO	126.5	97%	94%	No OMIM phenotype Intellectual disability (de Ligt (2012) N Eng J Med 367,1921)

				Autism (Sanders (2012) Nature 485,237)
TRIP12	132.7	99%	99%	No OMIM phenotype Autism (Iossifov (2012) Neuron 74,285)
TRMT10A	147.1	100%	100%	Microcephaly, short stature and impaired glucose metabolism, 616033
TSC1	127.2	99%	97%	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	111.2	99%	98%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN54	70.9	90%	77%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSPAN7	84.2	100%	100%	Mental retardation, X-linked 58, 300210
TTC8	88.4	98%	88%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTI2	92.8	99%	97%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	105.3	99%	93%	Lissencephaly 3, 611603
TUBA8	151.5	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB2B	88.7	100%	100%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB4A				Dystonia 4,torsion,autosomal dominant,128101 Leukodystrophy,hypomyelinating,612438
TUBGCP6	124.7	100%	97%	Microcephaly and chorioretinopathy, autosomal recessive 1, 251270
TUSC3	130	100%	98%	Mental retardation, autosomal recessive 7, 611093
TWIST1	104.6	97%	87%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400
UBE2A	62.6	96%	91%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	92.8	97%	92%	Angelman syndrome, 105830
UBE3B	121.1	100%	99%	Blepharophimosis-ptosis-intellectual disability syndrome, 615057
UBR1	130.1	99%	95%	Johanson-Blizzard syndrome, 243800
UPB1	151.8	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	35.5	88%	64%	Mental retardation, X-linked, syndromic 14, 300676

USP7	102.8	90%	83%	No OMIM phenotype Autism spectrum disorder (Levy (2011) Neuron 70,886)
USP9X	82.5	93%	84%	Mental retardation, X-linked 99, 300919
VLDLR	205.3	100%	98%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13B	140	98%	97%	Cohen syndrome, 216550
VRK1	127.4	100%	97%	Pontocerebellar hypoplasia type 1A, 607596
WAC	161.5	97%	92%	Desanto-Shinawi syndrome,616708
WDR13	70.1	99%	96%	No OMIM phenotype Intellectual disability,X-linked (Whibley (2010) Am J Hum Genet 87,173)
WDR19	133.5	100%	98%	Nephronophtisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR45	50.2	94%	89%	Neurodegeneration with brain iron accululation 5, 300894
WDR62	142.7	99%	97%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	123.3	100%	100%	Galloway-Mowat syndrome,251300
WDR81	135.5	99%	99%	Cerebellar ataxia, mental retardation and dysequilibrium syndrome 2, 610185
WWOX	122.5	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive, 12, 614322
XPA	46.6	95%	84%	Xeroderma pigmentosum, group A, 278700
XPNPEP3	128.2	97%	97%	Nephronophthisis-like nephropathy 1, 613159
XYLT1	124.1	87%	84%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	95.5	85%	79%	Coloboma, ocular with or without hearing impairment, cleft lip/palate and mental retardation, 120433
YWHAE	94.2	100%	93%	No OMIM phenotype Develomental delay,facial dysmorphology and growth retardation (Enomoto (2012) Am J Med Genet A 158A) Developmental delay and mild brain structural abnormalities (Bi (2009) Nat Genet 41,168)
YY1	138.3	100%	95%	No OMIM phenotype Mental retardation (Vissers (2010) Nat Genet 42,1109)
ZBTB16	154.8	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	189.6	99%	98%	?Mental retardation, autosomal dominant 22, 612337

ZDHHC15	58.2	99%	88%	?Mental retardation, X-linked 91, 300577
ZDHHC9	32.9	93%	74%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	149.6	100%	98%	Mowat-Wilson syndrome, 235730
ZFYVE26	110.3	99%	98%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC2	82.9	88%	76%	Holoprosencephaly-5, 609637
ZMYND11	125.8	100%	99%	Mental retardation, autosomal dominant 30, 616083
ZNF292	125.2	97%	96%	No OMIM phenotype Autism (Neale (2012) Nature 485,242)
ZNF41	81.1	100%	98%	Mental retardation, X-linked 89, 300848
ZNF592	109.2	99%	98%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF674	83.9	100%	100%	Mental retardation, X-linked 92, 300851
ZNF711	107.2	100%	96%	Mental retardation, X-linked 97, 300803
ZNF81	58.1	95%	89%	Mental retardation, X-linked 45, 300498

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 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