INTELLECTUAL DISABILITY GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABCC9	109,8	100%	99%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	58,2	73%	73%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	100,8	100%	99%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	116,3	100%	95%	Chanarin-Dorfman syndrome, 275630
ACAD9	90,8	100%	100%	ACAD9 deficiency, 611126
ACOX1	77,8	97%	94%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	81,9	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	127	100%	98%	Mental retardation, X-linked 63, 300387
ACTB	61,8	100%	95%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	60,2	97%	87%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACVR1	92,6	100%	98%	Fibrodysplasia ossificans progressiva, 135100
ACY1	78,5	100%	96%	Aminoacylase 1 deficiency, 609924
ADAR	131	99%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADCK3	105,5	100%	97%	Coenzyme Q10 deficiency, primary, 4, 612016
ADK	109,8	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300

ADSL	123,6	100%	99%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AFF2	136,4	99%	99%	Mental retardation, X-linked, FRAXE type, 309548
AGA	111,6	100%	91%	Aspartylglucosaminuria, 208400
AGPAT2	60,9	90%	85%	Lipodystrophy, congenital generalized, type 1, 608594
AGTR2	179,7	100%	100%	Mental retardation, X-linked 88, 300852
AHCY	76,9	91%	69%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	106,1	99%	98%	Joubert syndrome-3, 608629
AIFM1	111,7	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	117,9	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AK1	91,4	100%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AKT3	109,1	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALDH18A1	97,7	99%	95%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH3A2	91,7	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	71,8	93%	89%	Hyperprolinemia, type II, 239510
ALDH5A1	58,3	97%	89%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	45,6	45%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALX1	151,9	100%	100%	Frontonasal dysplasia 3, 613456
ALX4	72	100%	99%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMT	123,8	100%	100%	Glycine encephalopathy, 605899

ANKH	105,9	100%	100%	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKRD11	105,6	91%	86%	KBG syndrome, 148050
ANO10	106,7	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
AP1S2	164	100%	100%	Mental retardation, X-linked syndromic, Fried type, 300630
AP3B1	104,8	100%	99%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	102,2	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	121,8	100%	99%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	103,3	100%	99%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	82	100%	100%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	116,9	100%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	104,1	100%	99%	Periventricular heterotopia with microcephaly, 608097
ARG1	123,5	98%	90%	Argininemia, 207800
ARHGEF6	109,6	99%	98%	Mental retardation, X-linked 46, 300436
ARHGEF9	92,8	100%	97%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	102,4	99%	96%	Mental retardation, autosomal dominant 14, 614607
ARID1B	115,7	99%	97%	Mental retardation, autosomal dominant 12, 614562
ARL13B	124,4	100%	98%	Joubert syndrome 8, 612291

ARL6	134,4	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARX	60,5	80%	73%	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	72,8	97%	93%	Argininosuccinic aciduria, 207900
ASPA	105,1	100%	100%	Canavan disease, 271900
ASPM	134,8	100%	99%	Microcephaly 5, primary, autosomal recessive, 608716
ASXL1	142,1	98%	97%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ATIC	109,3	100%	97%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A2	100,9	100%	98%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481
ATP2A2	115,4	100%	100%	Darier disease, 124200 Acrokeratosis verruciformis, 101900
ATP6AP2	61,2	98%	91%	Mental retardation, X-linked, with epilepsy, 300423
ATP6V0A2	97,9	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	122,6	100%	100%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATR	113,1	100%	99%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564

ATRX	136,3	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	111,3	100%	100%	3-methylglutaconic aciduria, type I, 250950
B3GALTL	108,3	100%	96%	Peters-plus syndrome, 261540
B4GALT1	80,7	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	77,9	100%	96%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
BBS1	119,3	100%	99%	Bardet-Biedl syndrome 1, 209900
BBS10	121	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	143,2	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	113,4	100%	100%	Bardet-Biedl syndrome 2, 209900
BBS4	90	100%	97%	Bardet-Biedl syndrome 4, 209900
BBS5	130,3	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	119,3	100%	99%	Bardet-Biedl syndrome 7, 209900
BBS9	117,3	100%	100%	Bardet-Biedl syndrome 9, 209900
BCKDHA	100,9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	86,4	99%	83%	Maple syrup urine disease, type Ib, 248600
BCOR	125,9	100%	99%	Microphthalmia, syndromic 2, 300166
BCS1L	144	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BLM	114,8	100%	98%	Bloom syndrome, 210900

BRAF	72,6	100%	97%	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	115,6	99%	99%	Mental retardation, X-linked 93, 300659
BSCL2	104,1	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
BTD	132	100%	100%	Biotinidase deficiency, 253260
BUB1B	111,3	100%	99%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf57	68,3	100%	96%	Temtamy syndrome, 218340
C5ORF42	122,1	100%	99%	Joubert syndrome 17, 614615
CA2	146,1	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA8	79,5	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1C	97,2	98%	96%	Timothy syndrome, 601005 Brugada syndrome 3, 611875
CACNG2	106,8	100%	100%	Mental retardation, autosomal dominant 10, 614256
CASK	100,3	100%	100%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CBL	121,9	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	77,8	99%	83%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200

CC2D1A	91	100%	98%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	91,4	100%	98%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCBE1	83,8	95%	88%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC78	93	100%	100%	Myopathy, centronuclear, 4, 614807
CDH15	80,3	99%	95%	Mental retardation, autosomal dominant 3, 612580
CDK5RAP2	99,5	99%	96%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	129,7	100%	99%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CDON	109,8	99%	97%	Holoprosencephaly 11, 614226
CENPJ	127,2	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	118,3	99%	98%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	126,6	99%	99%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	93,6	99%	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	82,4	100%	100%	Joubert syndrome 15, 614464
CHD2	116,7	99%	98%	Epileptic encephalopathy, childhood-onset, 615369

CHD7	117,2	100%	99%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CLCNKB	75,4	87%	83%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLIC2	75,5	99%	94%	Mental retardation, X-linked, syndromic 32, 300886
CLN3	82,9	100%	98%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	125	97%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	72,1	98%	85%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	124,6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNTNAP2	100	100%	98%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COG1	120,4	99%	97%	Congenital disorder of glycosylation, type IIg, 611209
COG7	80,3	99%	94%	Congenital disorder of glycosylation, type IIe, 608779
COG8	108,6	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	82	98%	95%	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	80,5	100%	97%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COLEC11	112,7	100%	100%	3MC syndrome 2, 265050
COQ2	71,8	100%	88%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500

COX15	73	100%	98%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
CPS1	101,9	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CRBN	134	100%	100%	Mental retardation, autosomal recessive 2, 607417
CREBBP	78	99%	97%	Rubinstein-Taybi syndrome, 180849
CTDP1	71,2	89%	86%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNB1	113,8	99%	97%	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Pilomatricoma, somatic, 132600 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTSA	100,2	100%	99%	Galactosialidosis, 256540
CTSD	93,6	100%	97%	Ceroid lipofuscinosis, neuronal, 10, 610127
CUL4B	109,6	100%	99%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CYB5R3	83,3	100%	92%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
D2HGDH	61	98%	86%	D-2-hydroxyglutaric aciduria, 600721
DARS2	112,6	100%	99%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	104,9	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	95,3	100%	95%	Woodhouse-Sakati syndrome, 241080
DCX	115	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDHD2	99,1	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
DHCR24	81,2	99%	97%	Desmosterolosis, 602398

DHCR7	107	99%	98%	Smith-Lemli-Opitz syndrome, 270400
DHFR	50,6	79%	63%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	104,7	100%	99%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIP2B	100,2	99%	97%	Mental retardation, FRA12A type, 136630
DKC1	91,1	100%	98%	Dyskeratosis congenita, X-linked, 305000
DLD	131,2	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	75,7	97%	93%	Mental retardation, X-linked 90, 300850
DMD	107,4	100%	99%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DMPK	101,6	100%	96%	Myotonic dystrophy 1, 160900
DNAJC19	55,5	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNMT3B	91,6	100%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK8	83,9	100%	98%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DPAGT1	87,5	100%	94%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	145,4	100%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPYD	113,6	99%	97%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DST	157	100%	100%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DYM	93,4	100%	100%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326

DYM	93,4	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DYNC1H1	112,5	99%	97%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	135	99%	98%	Mental retardation, autosomal dominant 7, 614104
EFTUD2	86,7	98%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	93,8	96%	92%	Kleefstra syndrome, 610253
EIF2AK3	106,5	92%	91%	Wolcott-Rallison syndrome, 226980
ELOVL4	107,3	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMX2	106,9	100%	100%	Schizencephaly, 269160
EPB41L1	88,5	99%	95%	Mental retardation, autosomal dominant 11, 614257
ERCC2	85,9	99%	93%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	121,5	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC5	114,7	98%	98%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	141,3	98%	97%	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	88,4	100%	98%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	109,6	100%	98%	Spastic paraplegia 18, autosomal recessive, 611225

ESCO2	78	100%	99%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETHE1	57,6	100%	98%	Ethylmalonic encephalopathy, 602473
FAM126A	130,2	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FBN1	95,8	100%	98%	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	88,6	99%	95%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGFR1	115,2	99%	96%	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	116,7	100%	100%	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	66,6	94%	87%	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	85,3	96%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	80,2	99%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155

FKTN	112,9	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNA	119,2	100%	99%	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
FMR1	103,6	100%	100%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FOXG1	86,2	84%	78%	Rett syndrome, congenital variant, 613454
FOXP1	95,9	99%	95%	Mental retardation with language impairment and autistic features, 613670
FRAS1	95,6	98%	95%	Fraser syndrome, 219000
FTO	109,4	97%	97%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	86,2	92%	87%	Mental retardation, X-linked 9, 309549
FUCA1	80,4	100%	99%	Fucosidosis, 230000
GAD1	92,9	100%	99%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALE	110	100%	100%	Galactose epimerase deficiency, 230350
GALT	115,8	100%	100%	Galactosemia, 230400

GAMT	97,7	97%	92%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	109,9	100%	97%	Mental retardation, autosomal dominant 18, 615074
GATM	81,7	98%	89%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	96,1	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	14,5	52%	37%	Glycine encephalopathy, 605899
GDI1	139,8	100%	100%	Mental retardation, X-linked 41, 300849
GFAP	69,5	100%	95%	Alexander disease, 203450
GJB1	172,8	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	52,1	92%	82%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GK	48,3	90%	79%	Glycerol kinase deficiency, 307030
GLB1	77,2	98%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	57,6	97%	85%	Glycine encephalopathy, 605899
GLI2	104,9	100%	95%	Holoprosencephaly-9, 610829
GLI3	118	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	104,9	100%	100%	GM2-gangliosidosis, AB variant, 272750

GNAS	122,2	100%	99%	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	122	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNS	80,1	97%	88%	Mucopolysaccharidosis type IIID, 252940
GPC3	96	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	118,5	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GPR56	86,6	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854
GRIA3	101,9	100%	97%	Mental retardation, X-linked 94, 300699
GRIK2	109,2	100%	100%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	81,7	99%	96%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	134,7	100%	99%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	135,5	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRM1	138,5	100%	97%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSS	83,5	98%	97%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GTF2H5	89,9	100%	100%	Trichothiodystrophy, complementation group A, 601675
GUSB	67,7	89%	80%	Mucopolysaccharidosis VII, 253220

HAX1	129,3	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	107,1	100%	99%	Microphthalmia, syndromic 7, 309801
HCFC1	72,6	99%	94%	Mental retardation, X-linked 3, 309541
HDAC4	65,3	93%	90%	Brachydactyly-mental retardation syndrome, 600430

HDAC6	113,1	95%	94%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	98,1	100%	99%	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882
HESX1	94,7	100%	95%	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	91,9	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	105,5	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HLCS	140,4	100%	100%	Holocarboxylase synthetase deficiency, 253270
HOXA1	123,3	100%	100%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HPD	101,1	100%	98%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	96,2	100%	98%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HRAS	96,1	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	105,3	99%	92%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSPD1	16,8	59%	40%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HUWE1	95,7	99%	97%	Mental retardation, X-linked syndromic, Turner type, 300706
IDS	101,3	89%	85%	Mucopolysaccharidosis II, 309900
IDUA	81	95%	89%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IER3IP1	58,5	100%	93%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IGBP1	93,4	94%	88%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	124,4	100%	96%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	25,9	26%	26%	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	135,7	100%	100%	Mental retardation, X-linked 21/34, 300143
INPP5E	68,4	98%	94%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
IQSEC2	79,7	95%	87%	Mental retardation, X-linked 1, 309530
ISPD	83,8	97%	94%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
JAM3	72,5	95%	90%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	35,1	79%	64%	Koolen-De Vries syndrome, 610443

KAT6B	140,2	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNJ10	148,5	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	127,4	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		0%	0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	71,9	100%	98%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KCNT1	72	100%	96%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCTD7	122,9	91%	84%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	113	100%	100%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 -3
KDM6A	120	100%	99%	Kabuki syndrome 2, 300867
KIAA1279	97,7	99%	97%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF11	95,9	100%	98%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	69	99%	92%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF7	65	93%	82%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	78,5	99%	95%	Mental retardation, autosomal dominant 4, 612581

KRAS	70,6	97%	90%	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myelogenous Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Breast cancer, somatic, 114480 SFM syndrome, somatic mosaic, 163200
L1CAM	121,7	100%	99%	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	78,2	100%	96%	L-2-hydroxyglutaric aciduria, 236792
LAMA2	96,4	99%	97%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMC3	93,4	100%	93%	Cortical malformations, occipital, 614115
LAMP2	112,8	100%	98%	Danon disease, 300257
LARGE	97	97%	92%	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	99,8	100%	100%	Alazami syndrome, 615071
LIG4	177,2	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LRP2	104,6	100%	98%	Donnai-Barrow syndrome, 222448
LRPPRC	92,4	100%	97%	Leigh syndrome, French-Canadian type, 220111
MAGT1	97,6	100%	100%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853

MAN1B1	96,6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	81,3	99%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,6	100%	99%	Mannosidosis, beta, 248510
MAOA	104,8	100%	100%	Brunner syndrome, 300615
MAP2K1	95,9	99%	84%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	111,8	100%	98%	Cardiofaciocutaneous syndrome 4, 615280
MAT1A	90	100%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	140	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	126,7	100%	100%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MCCC1	94,6	100%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	105,2	95%	89%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	101,3	97%	93%	Mucolipidosis IV, 252650
MCPH1	118,2	100%	100%	genetic heterogeneity Microcephaly 1, primary, autosomal recessive, 251200
MECP2	165,6	100%	99%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830 Mental retardation, X-linked syndromic, Lubs type, 300260
MED12	122,4	97%	94%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895

MED17	138,3	99%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	108,6	100%	99%	Mental retardation, autosomal recessive 18, 614249
MEF2C	104,5	100%	99%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443
MGAT2	194,6	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MID1	146,4	100%	99%	Opitz GBBB syndrome, type I, 300000
MKKS	137,9	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MLYCD	69,5	89%	84%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	160,5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMACHC	175,4	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	68,9	100%	100%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MOCS1	76,3	99%	95%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	103	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MPDU1	117,6	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPLKIP	66	100%	100%	Trichothiodystrophy, nonphotosensitive 1, 234050
MRPS22	96,7	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MTR	100,3	99%	98%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	102,8	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	115,5	100%	100%	Methylmalonic aciduria, mut(0) type, 251000

MVK	86,1	100%	99%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYCN	95	99%	97%	Feingold syndrome, 164280
MYO5A	91,5	99%	97%	Griscelli syndrome, type 1, 214450
NAA10	95,4	97%	96%	N-terminal acetyltransferase deficiency, 300855
NAGA	82,1	100%	95%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	67	94%	84%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	105,1	99%	96%	?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419
NDE1	95,3	100%	97%	Lissencephaly 4 (with microcephaly), 614019
NDP	79,4	99%	94%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NDUFA1	189,6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	98,8	98%	82%	Mitochondrial complex I deficiency, 252010
NDUFA12	89,4	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFS1	76,8	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	122	100%	96%	Mitochondrial complex I deficiency, 252010
NDUFS3	149,1	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	126,6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS7	112,5	100%	100%	Leigh syndrome, 256000
NDUFS8	105,4	100%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000

NDUFV1	62,3	97%	91%	Mitochondrial complex I deficiency, 252010
NEU1	17,2	61%	35%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NF1	79	83%	81%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Melanoma, desmoplastic neurotrophic (2) Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NHS	126,9	96%	92%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIPBL	113,7	99%	98%	Cornelia de Lange syndrome 1, 122470
NKX2-1	98,4	100%	98%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLRP3	116,6	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
NPHP1	110,7	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NRAS	135	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	118,7	99%	98%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	119,4	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
NSDHL	100	99%	97%	CHILD syndrome, 308050 CK syndrome, 300831

NSUN2	122,3	100%	92%	Mental retardation, autosomal recessive 5, 611091
NTRK1	65	98%	89%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
OCLN	91,1	72%	71%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	111,5	98%	97%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	67,8	93%	88%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
OPHN1	98,8	99%	98%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	103,5	100%	97%	Meier-Gorlin syndrome 1, 224690
OTC	103,3	100%	99%	CGD Ornithine transcarbamylase deficiency, 311250
PACS1	99,9	97%	95%	Mental retardation, autosomal dominant 17, 615009
PAH	82,4	100%	95%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	100,9	100%	100%	Mental retardation, X-linked 30/47, 300558
PANK2	113,2	99%	93%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX6	91,4	100%	98%	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	65,9	100%	90%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700

PC	96,5	97%	91%	Pyruvate carboxylase deficiency, 266150
PCDH19	138,2	99%	98%	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	92	97%	90%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 -3
PDHA1	113,8	100%	99%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDSS1	94,9	88%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	92,3	100%	98%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	63,9	100%	90%	Prolidase deficiency, 170100
PEX1	118,6	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73,1	89%	85%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	167,6	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	114	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	134,7	98%	96%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	92,5	94%	90%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	107,5	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	150,5	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	111,4	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	130,7	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	86	98%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370

PEX6	89,8	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	98,2	99%	89%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGAP2	134,7	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	76,4	85%	77%	Phosphoglycerate kinase 1 deficiency, 300653
PHF6	135,5	100%	100%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	100,2	100%	99%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	85,1	100%	99%	Phosphoglycerate dehydrogenase deficiency, 601815
PIGN	102,3	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	104,9	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGV	165	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3R2	76,5	90%	83%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PLA2G6	74,8	100%	92%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLCB1	107,8	99%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	77,9	100%	94%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	85,6	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	67,7	99%	95%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	117,2	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POC1A	101,6	98%	94%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813

POLR3A	85,6	99%	95%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100,3	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	99,5	100%	98%	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	98,2	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	69,5	98%	92%	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	113,4	92%	90%	Focal dermal hypoplasia, 305600
PPOX	92,4	100%	97%	Porphyria variegata, 176200
PPT1	68,4	100%	94%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	124,6	100%	100%	Renpenning syndrome, 309500
PRODH	52	78%	63%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	131,1	100%	100%	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	97,6	100%	95%	Mental retardation, autosomal recessive 1, 249500
PSAP	83,5	100%	99%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722

PTCH1	82,8	99%	95%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTEN	122,4	99%	94%	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	41,7	83%	68%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PUS1	65,6	99%	96%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PVRL1	76,9	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PYCR1	86,8	100%	98%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB18	105,6	100%	100%	Warburg micro syndrome 3, 614222
RAB27A	119,7	100%	100%	Griscelli syndrome, type 2, 607624
RAB39B	166,5	100%	100%	Mental retardation, X-linked 72, 300271
RAB3GAP1	124,9	100%	98%	Warburg micro syndrome 1, 600118
RAB3GAP2	106,7	100%	98%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225

RAB40AL	29,8	99%	82%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519 -3
RAD21	86,4	98%	90%	Cornelia de Lange syndrome 4, 614701
RAF1	89,7	100%	98%	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAI1	133,7	99%	98%	Immunodeficiency 9, 612782
RAI1	133,7	99%	98%	Smith-Magenis syndrome, 182290
RARS2	82,3	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RBM10	99,9	100%	98%	TARP syndrome, 311900
RBM28	104,4	100%	98%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RELN	102,7	99%	97%	Lissencephaly 2 (Norman-Roberts type), 257320
RFT1	73,9	100%	96%	Congenital disorder of glycosylation, type In, 612015
RIT1	134,2	100%	100%	Noonan syndrome 8, 615355
RMND1	69,7	91%	89%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH2A	96,1	99%	94%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,5	99%	97%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	133	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	85	100%	99%	Leukoencephalopathy, cystic, without megalencephaly, 612951
ROGDI	94,2	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPGRIP1L	94,2	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RPS6KA3	97,9	100%	99%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844

SALL1	137,1	100%	99%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SATB2	100,2	100%	96%	Cleft palate and mental retardation, 119540
SC5D	146,4	100%	100%	Lathosterolosis, 607330
SCN1A	114,5	100%	99%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403
SCN2A	121,7	99%	99%	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721
SCN8A	134,4	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCO2	82,5	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	8,6	34%	12%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SERAC1	88,8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	141,9	98%	96%	Schinzel-Giedion midface retraction syndrome, 269150
SHANK3	68,6	90%	80%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	94	100%	91%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SHOC2	116,4	100%	98%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	129,2	100%	99%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIL1	97,3	100%	100%	Marinesco-Sjogren syndrome, 248800

SIX3	115,4	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SKI	63,3	89%	79%	Shprintzen-Goldberg syndrome, 182212
SLC12A6	88,7	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy, 218000 -3
SLC16A2	93,9	100%	99%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	95,4	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC25A15	102,9	95%	80%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC25A22	73,5	98%	93%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	88,3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC33A1	92,1	100%	99%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35C1	97,4	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC4A4	98,9	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A8	7,8	20%	11%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	110,6	100%	97%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	123,6	100%	98%	Pancreatic cancer Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210
SMARCA2	86,3	97%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	83,3	98%	92%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609

SMARCB1	116	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608
SMC3	107,1	98%	97%	Cornelia de Lange syndrome 3, 610759
SMOC1	73,4	100%	94%	Microphthalmia with limb anomalies, 206920
SMPD1	103,9	97%	90%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	26,3	87%	62%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP29	118	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SOBP	106	98%	92%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOS1	113,2	100%	99%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SOX10	67,6	100%	100%	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136
SOX3	83,3	98%	95%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SPG11	104,6	99%	97%	Spastic paraplegia 11, autosomal recessive, 604360
SPRED1	129,3	100%	100%	Legius syndrome, 611431
SPTAN1	95,4	99%	97%	Epileptic encephalopathy, early infantile, 5, 613477
SRCAP	133,4	100%	99%	Floating-Harbor syndrome, 136140
SRD5A3	122,5	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	81,6	100%	97%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 -3
ST3GAL3	120,3	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006

STIL	136,7	100%	99%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	66,6	99%	94%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STXBP1	92,5	100%	97%	Epileptic encephalopathy, early infantile, 4, 612164 (2)
SUOX	174,6	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	86,7	88%	88%	Leigh syndrome, due to COX deficiency, 256000
SYN1	69,1	96%	75%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	52,9	93%	81%	Mental retardation, autosomal dominant 5, 612621
SYP	93,4	100%	99%	Mental retardation, X-linked 96, 300802
SYT14	121,6	94%	90%	Spinocerebellar ataxia, autosomal recessive 11, 614229
TAT	92	100%	98%	Tyrosinemia, type II, 276600
TBC1D24	109,5	100%	100%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TBCE	117,6	100%	100%	Kenny-Caffey syndrome-1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410
TCF4	94	100%	99%	Pitt-Hopkins syndrome, 610954
TECR	83,1	100%	91%	Mental retardation, autosomal recessive 14, 614020
TGFBR1	117,7	95%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800

TGFBR2	86,2	100%	97%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
THRB	114,6	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	58	94%	84%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMCO1	76,6	100%	98%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM165	86,6	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	73,4	96%	92%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397 -3
TMEM237	89,6	100%	99%	Joubert syndrome 14, 614424
TMEM67	110,2	100%	100%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TPP1	119,2	100%	96%	Ceroid lipofuscinosis, neuronal, 2, 204500
TRAPPC9	67,4	98%	94%	Mental retardation, autosomal recessive 13, 613192
TREX1	134,4	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TSC1	89	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	81,7	98%	95%	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690

TSPAN7	86,4	99%	95%	Mental retardation, X-linked 58, 300210
TTC8	103,3	100%	99%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TUBA1A	23,5	89%	51%	Lissencephaly 3, 611603
TUBA8	95	99%	97%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB2B	54,7	100%	97%	Polymicrogyria, symmetric or asymmetric, 610031
TUSC3	118,3	100%	100%	Mental retardation, autosomal recessive 7, 611093
UBE2A	101,2	100%	100%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	102,1	100%	99%	Angelman syndrome, 105830
UBR1	101	100%	100%	Johanson-Blizzard syndrome, 243800
UPB1	114,7	100%	97%	Beta-ureidopropionase deficiency, 613161
UPF3B	98,8	100%	95%	Mental retardation, X-linked, syndromic 14, 300676
VLDLR	106,1	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13B	104,8	100%	99%	Cohen syndrome, 216550
VRK1	124,6	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
WDR45	91,6	100%	99%	Neurodegeneration with brain iron acculation 5, 300894
WDR62	103,8	99%	94%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
XPA	80,9	100%	93%	Xeroderma pigmentosum, group A, 278700
XPNPEP3	119	100%	98%	Nephronophthisis-like nephropathy 1, 613159
ZBTB16	115,8	100%	99%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447

ZDHHC9	92,5	100%	99%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	152,6	100%	99%	Mowat-Wilson syndrome, 235730
ZIC2	62,3	93%	87%	Holoprosencephaly-5, 609637
ZNF41	117,8	100%	100%	Mental retardation, X-linked 89, 300848
ZNF592	107,3	93%	91%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF674	123,9	100%	96%	Mental retardation, X-linked 92, 300851
ZNF711	134,6	100%	100%	Mental retardation, X-linked 97, 300803
ZNF81	108,5	100%	99%	Mental retardation, X-linked 45, 300498

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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 : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors