

TABLE S2. ID gene panel listing 490 candidate genes for ID

Gene	Chr	Omim disease
<i>ADAR</i>	1	Aicardi-Goutieres syndrome 6
<i>ADCK3</i>	1	-
<i>AKT3</i>	1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome
<i>ALG6</i>	1	Congenital disorder of glycosylation type Ic
<i>AP4B1</i>	1	Spastic paraplegia 47 autosomal recessive
<i>ARID1A</i>	1	Mental retardation autosomal dominant 14
<i>ASPM</i>	1	Microcephaly 5 primary autosomal recessive
<i>ATP1A2</i>	1	Alternating hemiplegia of childhood
<i>DARS2</i>	1	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
<i>DBT</i>	1	Maple syrup urine disease type II
<i>DHCR24</i>	1	Desmosterolosis
<i>DPYD</i>	1	5-fluorouracil toxicity
<i>FH</i>	1	Fumarase deficiency
<i>FUCA1</i>	1	Fucosidosis
<i>GALE</i>	1	Galactose epimerase deficiency
<i>GATAD2B</i>	1	Mental retardation autosomal dominant 18
<i>GJC2</i>	1	Leukodystrophy hypomyelinating 2
<i>GNPAT</i>	1	Chondrodysplasia punctata rhizomelic type 2
<i>HAX1</i>	1	Neutropenia severe congenital 3 autosomal recessive
<i>MMACHC</i>	1	Methylmalonic aciduria and homocystinuria cblC type
<i>MTR</i>	1	Homocystinuria-megaloblastic anemia cblG complementation type
<i>NDUFS2</i>	1	Mitochondrial complex I deficiency
<i>NLRP3</i>	1	CINCA syndrome
<i>NTRK1</i>	1	Insensitivity to pain congenital with anhidrosis

<i>ORC1</i>	1	Meier-Gorlin syndrome 1
<i>PEX10</i>	1	Peroxisome biogenesis disorder 6A (Zellweger)
<i>PEX11B</i>	1	Peroxisome biogenesis disorder 14B
<i>PHGDH</i>	1	Phosphoglycerate dehydrogenase deficiency
<i>PIGV</i>	1	Hyperphosphatasia with mental retardation syndrome 1
<i>POMGNT1</i>	1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3
<i>PPOX</i>	1	Porphyria variegata
<i>RAB3GAP2</i>	1	Martsof syndrome
<i>SKI</i>	1	Shprintzen-Goldberg syndrome
<i>SLC2A1</i>	1	Dystonia 9
<i>ST3GAL3</i>	1	Epileptic encephalopathy early infantile 15
<i>STIL</i>	1	Microcephaly 7 primary autosomal recessive
<i>SYT14</i>	1	Spinocerebellar ataxia autosomal recessive 11
<i>TBCE</i>	1	Hypoparathyroidism-retardation-dysmorphism syndrome
<i>TMCO1</i>	1	Craniofacial dysmorphism skeletal anomalies and mental retardation syndrome
<i>ACVR1</i>	2	Fibrodysplasia ossificans progressiva
<i>BBS5</i>	2	Bardet-Biedl syndrome 5
<i>BCS1L</i>	2	Bjornstad syndrome
<i>COLEC11</i>	2	3MC syndrome 2
<i>D2HGDH</i>	2	D-2-hydroxyglutaric aciduria
<i>EIF2AK3</i>	2	Wolcott-Rallison syndrome
<i>ERCC3</i>	2	Trichothiodystrophy
<i>GAD1</i>	2	Cerebral palsy spastic quadriplegic 1
<i>GLI2</i>	2	Holoprosencephaly-9
<i>HDAC4</i>	2	Brachydactyly-mental retardation syndrome
<i>LRP2</i>	2	Donnai-Barrow syndrome
<i>LRPPRC</i>	2	Leigh syndrome French-Canadian type
<i>MBD5</i>	2	Mental retardation autosomal dominant 1
<i>MMADHC</i>	2	-
<i>MYCN</i>	2	Feingold syndrome
<i>NDUFS1</i>	2	Mitochondrial complex I deficiency

<i>NPHP1</i>	2	Joubert syndrome 4
<i>NRXN1</i>	2	Pitt-Hopkins-like syndrome 2
<i>PEX13</i>	2	Peroxisome biogenesis disorder 11A (Zellweger)
<i>RAB3GAP1</i>	2	Warburg micro syndrome 1
<i>SATB2</i>	2	Cleft palate and mental retardation
<i>SCN1A</i>	2	Dravet syndrome
<i>SCN2A</i>	2	Epileptic encephalopathy early infantile 11
<i>SIX3</i>	2	Holoprosencephaly-2
<i>SOS1</i>	2	Fibromatosis gingival
<i>TMEM237</i>	2	Joubert syndrome 14
<i>ZEB2</i>	2	Mowat-Wilson syndrome
<i>ABHD5</i>	3	Chanarin-Dorfman syndrome
<i>ACAD9</i>	3	ACAD9 deficiency
<i>ALG3</i>	3	Congenital disorder of glycosylation type Id
<i>AMT</i>	3	Glycine encephalopathy
<i>ANO10</i>	3	Spinocerebellar ataxia autosomal recessive 10
<i>ARL13B</i>	3	Joubert syndrome 8
<i>ARL6</i>	3	Bardet-Biedl syndrome 3
<i>ATR</i>	3	Cutaneous telangiectasia and cancer syndrome familial
<i>CRBN</i>	3	Mental retardation autosomal recessive 2
<i>CTNNB1</i>	3	Colorectal cancer somatic
<i>DNAJC19</i>	3	3-methylglutaconic aciduria type V
<i>FANCD2</i>	3	Fanconi anemia complementation group D2
<i>FOXP1</i>	3	Mental retardation with language impairment and autistic features
<i>HESX1</i>	3	Growth hormone deficiency with pituitary anomalies
<i>MCCC1</i>	3	3-Methylcrotonyl-CoA carboxylase 1 deficiency
<i>MRPS22</i>	3	Combined oxidative phosphorylation deficiency 5
<i>POC1A</i>	3	Short stature onychodysplasia facial dysmorphism and hypotrichosis
<i>RAF1</i>	3	LEOPARD syndrome 2
<i>RFT1</i>	3	Congenital disorder of glycosylation type In
<i>SLC33A1</i>	3	Congenital cataracts hearing loss and neurodegeneration
<i>SOX2-OT</i>	3	-

<i>TGFBR2</i>	3	Colorectal cancer hereditary nonpolyposis type 6
<i>THRB</i>	3	Thyroid hormone resistance
<i>TREX1</i>	3	Aicardi-Goutieres syndrome 1 dominant and recessive
<i>AGA</i>	4	Aspartylglucosaminuria
<i>AIMP1</i>	4	Leukodystrophy hypomyelinating 3
<i>BBS12</i>	4	Bardet-Biedl syndrome 12
<i>BBS7</i>	4	Bardet-Biedl syndrome 7
<i>CC2D2A</i>	4	COACH syndrome
<i>CEP135</i>	4	Microcephaly 8 primary autosomal recessive
<i>COQ2</i>	4	Coenzyme Q10 deficiency primary 1
<i>FGFR3</i>	4	Achondroplasia
<i>FRAS1</i>	4	Fraser syndrome
<i>IDUA</i>	4	Mucopolysaccharidosis Ih
<i>LARP7</i>	4	Alazami syndrome
<i>MANBA</i>	4	Mannosidosis beta
<i>MMAA</i>	4	Methylmalonic aciduria vitamin B12-responsive
<i>PRSS12</i>	4	Mental retardation autosomal recessive 1
<i>SLC4A4</i>	4	Renal tubular acidosis proximal with ocular abnormalities
<i>SRD5A3</i>	4	Congenital disorder of glycosylation type Iq
<i>TMEM165</i>	4	Congenital disorder of glycosylation type IIk
<i>ANKH</i>	5	Chondrocalcinosis 2
<i>AP3B1</i>	5	Hermansky-Pudlak syndrome 2
<i>B4GALT7</i>	5	Ehlers-Danlos syndrome progeroid type 1
<i>C5orf42</i>	5	Joubert syndrome 17
<i>DHFR</i>	5	Megaloblastic anemia due to dihydrofolate reductase deficiency
<i>ERCC8</i>	5	Cockayne syndrome type A
<i>MCCC2</i>	5	3-Methylcrotonyl-CoA carboxylase 2 deficiency Mental retardation stereotypic movements epilepsy and/or cerebral
<i>MEF2C</i>	5	malformations
<i>MOCS2</i>	5	Molybdenum cofactor deficiency type B
<i>MTRR</i>	5	Homocystinuria-megaloblastic anemia cbl E type
<i>NDUFS4</i>	5	Leigh syndrome

<i>NIPBL</i>	5	Cornelia de Lange syndrome 1
<i>NSD1</i>	5	Beckwith-Wiedemann syndrome
<i>NSUN2</i>	5	Mental retardation autosomal recessive 5
<i>OCLN</i>	5	Band-like calcification with simplified gyration and polymicrogyria
<i>SDHA</i>	5	Cardiomyopathy dilated 1GG
<i>SIL1</i>	5	Marinesco-Sjogren syndrome
<i>AHI1</i>	6	Joubert syndrome-3
<i>ALDH5A1</i>	6	Succinic semialdehyde dehydrogenase deficiency
<i>ARID1B</i>	6	Mental retardation autosomal dominant 12
<i>BCKDHB</i>	6	Maple syrup urine disease type 1b
<i>ELOVL4</i>	6	Ichthyosis spastic quadriplegia and mental retardation
<i>GRIK2</i>	6	Mental retardation autosomal recessive 6
<i>GTF2H5</i>	6	Trichothiodystrophy complementation group A
<i>LAMA2</i>	6	Muscular dystrophy congenital merosin-deficient
<i>MED23</i>	6	Mental retardation autosomal recessive 18
<i>MOCS1</i>	6	Molybdenum cofactor deficiency type A
<i>MUT</i>	6	Methylmalonic aciduria mut(0) type
<i>NEU1</i>	6	Sialidosis type I
<i>PDSS2</i>	6	Coenzyme Q10 deficiency primary 3
<i>PEX7</i>	6	Peroxisome biogenesis disorder 9B
<i>RARS2</i>	6	Pontocerebellar hypoplasia type 6
<i>RMND1</i>	6	Combined oxidative phosphorylation deficiency 11
<i>SERAC1</i>	6	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome
<i>SLC17A5</i>	6	Salla disease
<i>SOBP</i>	6	Mental retardation anterior maxillary protrusion and strabismus
<i>SYNGAP1</i>	6	Mental retardation autosomal dominant 5
<i>TUBB2B</i>	6	Polymicrogyria symmetric or asymmetric
<i>ACTB</i>	7	Baraitser-Winter syndrome 1
<i>ASL</i>	7	Argininosuccinic aciduria
<i>BBS9</i>	7	Bardet-Biedl syndrome 9
<i>BRAF</i>	7	Adenocarcinoma of lung somatic

<i>CEP41</i>	7	Joubert syndrome 15
<i>CNTNAP2</i>	7	Cortical dysplasia-focal epilepsy syndrome
<i>DLD</i>	7	Dihydrolipoamide dehydrogenase deficiency
<i>GLI3</i>	7	Greig cephalopolysyndactyly syndrome
<i>GUSB</i>	7	Mucopolysaccharidosis VII
<i>HOXA1</i>	7	Athabaskan brainstem dysgenesis syndrome
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye
<i>ISPD</i>	7	anomalies) type A 7
<i>KCTD7</i>	7	Epilepsy progressive myoclonic 3 with or without intracellular inclusions
<i>PEX1</i>	7	Peroxisome biogenesis disorder 1A (Zellweger)
<i>RBM28</i>	7	Alopecia neurologic defects and endocrinopathy syndrome
<i>RELN</i>	7	Lissencephaly 2 (Norman-Roberts type)
<i>SHH</i>	7	Holoprosencephaly-3
<i>CA2</i>	8	Osteopetrosis autosomal recessive 3 with renal tubular acidosis
<i>CHD7</i>	8	CHARGE syndrome
<i>DDHD2</i>	8	Spastic paraplegia 54 autosomal recessive
<i>ERLIN2</i>	8	Spastic paraplegia 18 autosomal recessive
<i>ESCO2</i>	8	Roberts syndrome
<i>KCNK9</i>	8	Birk-Barel mental retardation dysmorphism syndrome
<i>MCPH1</i>	8	Microcephaly 1 primary autosomal recessive
<i>NBN</i>	8	Leukemia, acute lymphoblastic
<i>RAD21</i>	8	Cornelia de Lange syndrome 4
<i>TMEM67</i>	8	COACH syndrome
<i>TRAPPC9</i>	8	Mental retardation autosomal recessive 13
<i>TUSC3</i>	8	Mental retardation autosomal recessive 7
<i>VPS13B</i>	8	Cohen syndrome
<i>AGPAT2</i>	9	Lipodystrophy congenital generalized type 1
<i>AK1</i>	9	Hemolytic anemia due to adenylate kinase deficiency
<i>ALG2</i>	9	Congenital disorder of glycosylation type II
<i>APTX</i>	9	Ataxia early-onset with oculomotor apraxia and hypoalbuminemia
<i>AUH</i>	9	3-methylglutaconic aciduria type I
<i>B4GALT1</i>	9	Congenital disorder of glycosylation type IIb

<i>CDK5RAP2</i>	9	Microcephaly 3 primary autosomal recessive
<i>DOCK8</i>	9	Hyper-IgE recurrent infection syndrome autosomal recessive
<i>EHMT1</i>	9	Kleefstra syndrome
<i>FKTN</i>	9	Cardiomyopathy dilated 1X
<i>GALT</i>	9	Galactosemia
<i>GLDC</i>	9	Glycine encephalopathy
<i>GRIN1</i>	9	Mental retardation autosomal dominant 8
<i>INPP5E</i>	9	Joubert syndrome 1
<i>KCNT1</i>	9	Epilepsy nocturnal frontal lobe 5
<i>LAMC3</i>	9	Cortical malformations occipital
<i>MAN1B1</i>	9	Mental retardation autosomal recessive 15
<i>PIGO</i>	9	Hyperphosphatasia with mental retardation syndrome 2
<i>POMT1</i>	9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1
<i>PTCH1</i>	9	Basal cell carcinoma somatic
<i>SMARCA2</i>	9	Nicolaides-Baraitser syndrome
<i>STXBP1</i>	9	Epileptic encephalopathy early infantile 4
<i>SURF1</i>	9	Leigh syndrome due to COX deficiency
<i>TGFBR1</i>	9	Loeys-Dietz syndrome type 1A
<i>TSC1</i>	9	Focal cortical dysplasia Taylor balloon cell type
<i>VLDLR</i>	9	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1
<i>XPA</i>	9	Xeroderma pigmentosum group A
<i>ALDH18A1</i>	10	Cutis laxa autosomal recessive type IIIA
<i>COX15</i>	10	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 2
<i>DHTKD1</i>	10	2-aminoadipic 2-oxoadipic aciduria
<i>EMX2</i>	10	Schizencephaly
<i>ERCC6</i>	10	Cerebrooculofacioskeletal syndrome 1
<i>FGFR2</i>	10	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
<i>KAT6B</i>	10	Genitopatellar syndrome
<i>KIAA1279</i>	10	Goldberg-Shprintzen megacolon syndrome

<i>KIF11</i>	10	Microcephaly with or without chorioretinopathy lymphedema or mental retardation Hypermethioninemia persistent autosomal dominant due to methionine
<i>MAT1A</i>	10	adenosyltransferase I/III deficiency
<i>PDSS1</i>	10	Coenzyme Q10 deficiency primary 2
<i>POLR3A</i>	10	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism
<i>PTEN</i>	10	Bannayan-Riley-Ruvalcaba syndrome
<i>RAB18</i>	10	Warburg micro syndrome 3
<i>SHOC2</i>	10	Noonan-like syndrome with loose anagen hair
<i>SMC3</i>	10	-
<i>ALG9</i>	11	Congenital disorder of glycosylation type II
<i>BBS1</i>	11	Bardet-Biedl syndrome 1
<i>BSCL2</i>	11	Lipodystrophy congenital generalized type 2
<i>CDON</i>	11	Holoprosencephaly 11
<i>DHCR7</i>	11	Smith-Lemli-Opitz syndrome
<i>DPAGT1</i>	11	Congenital disorder of glycosylation type Ij
<i>HRAS</i>	11	Congenital myopathy with excess of muscle spindles
<i>JAM3</i>	11	Hemorrhagic destruction of the brain subependymal calcification and cataracts
<i>KCNJ11</i>	11	Diabetes mellitus permanent neonatal with neurologic features
<i>KIRREL3</i>	11	Mental retardation autosomal dominant 4
<i>MED17</i>	11	Microcephaly postnatal progressive with seizures and brain atrophy
<i>NDUFS3</i>	11	Leigh syndrome due to mitochondrial complex I deficiency
<i>NDUFS8</i>	11	Leigh syndrome due to mitochondrial complex I deficiency
<i>NDUFV1</i>	11	Mitochondrial complex I deficiency
<i>PACS1</i>	11	Mental retardation autosomal dominant 17
<i>PAX6</i>	11	Aniridia
<i>PC</i>	11	Pyruvate carboxylase deficiency
<i>PVRL1</i>	11	-
<i>RNASEH2C</i>	11	Aicardi-Goutieres syndrome 3
<i>SLC25A22</i>	11	Epileptic encephalopathy early infantile 3
<i>SLC35C1</i>	11	Congenital disorder of glycosylation type IIc

<i>SMPD1</i>	11	Niemann-Pick disease type A
<i>ABCC9</i>	12	Atrial fibrillation familial 12
<i>ATP2A2</i>	12	Acrokeratosis verruciformis
<i>ATP6V0A2</i>	12	Cutis laxa autosomal recessive type IIA
<i>BBS10</i>	12	Bardet-Biedl syndrome 10
<i>CACNA1C</i>	12	Brugada syndrome 3
<i>CEP290</i>	12	Bardet-Biedl syndrome 14
<i>DIP2B</i>	12	Mental retardation FRA12A type
<i>GNS</i>	12	Mucopolysaccharidosis type IIID
<i>GRIN2B</i>	12	Mental retardation autosomal dominant 6
<i>HPD</i>	12	Hawkinsinuria
<i>IGF1</i>	12	Growth retardation with deafness and mental retardation due to IGF1 deficiency
<i>KRAS</i>	12	Bladder cancer somatic
<i>MVK</i>	12	Hyper-IgD syndrome
<i>NDUFA12</i>	12	Leigh syndrome due to mitochondrial complex 1 deficiency
<i>PEX5</i>	12	Peroxisome biogenesis disorder 2A (Zellweger)
<i>POLR3B</i>	12	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism
<i>PTPN11</i>	12	LEOPARD syndrome 1
<i>PUS1</i>	12	Mitochondrial myopathy and sideroblastic anemia 1
<i>SCN8A</i>	12	Cognitive impairment with or without cerebellar ataxia
<i>SUOX</i>	12	Sulfite oxidase deficiency
<i>TUBA1A</i>	12	Lissencephaly 3
<i>B3GALTL</i>	13	Peters-plus syndrome
<i>BIVM-</i>	13	-
<i>ERCC5</i>		
<i>CENPJ</i>	13	Microcephaly 6 primary autosomal recessive
<i>COL4A1</i>	13	Angiopathy hereditary with nephropathy aneurysms and muscle
<i>COL4A2</i>	13	Porencephaly 2
<i>LIG4</i>	13	LIG4 syndrome
<i>RNASEH2B</i>	13	Aicardi-Goutieres syndrome 2
<i>SLC25A15</i>	13	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome

<i>ZIC2</i>	13	Holoprosencephaly-5
<i>ABCD4</i>	14	Methylmalonic aciduria and homocystinuria cblJ type
<i>AP4S1</i>	14	Spastic paraplegia 52 autosomal recessive
<i>DYNC1H1</i>	14	Charcot-Marie-Tooth disease axonal type 20
<i>FOXP1</i>	14	Rett syndrome congenital variant
<i>GCH1</i>	14	Dystonia DOPA-responsive with or without hyperphenylalaninemia
<i>GPHN</i>	14	Molybdenum cofactor deficiency type C
<i>L2HGDH</i>	14	L-2-hydroxyglutaric aciduria
<i>MGAT2</i>	14	Congenital disorder of glycosylation type IIa
<i>NKX2-1</i>	14	Chorea hereditary benign
<i>PNP</i>	14	Immunodeficiency due to purine nucleoside phosphorylase deficiency
<i>POMT2</i>	14	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2
<i>SMOC1</i>	14	Microphthalmia with limb anomalies
<i>TTC8</i>	14	Bardet-Biedl syndrome 8
<i>AP4E1</i>	15	Spastic paraplegia 51 autosomal recessive
<i>BBS4</i>	15	Bardet-Biedl syndrome 4
<i>BLM</i>	15	Bloom syndrome
<i>BUB1B</i>	15	Colorectal cancer somatic
<i>CEP152</i>	15	Microcephaly 9 primary autosomal recessive
<i>FBN1</i>	15	Acromicric dysplasia
<i>GATM</i>	15	Cerebral creatine deficiency syndrome 3
<i>KIF7</i>	15	Acrocallosal syndrome
<i>MAP2K1</i>	15	Cardiofaciocutaneous syndrome 3
<i>MYO5A</i>	15	Griscelli syndrome type 1
<i>RAB27A</i>	15	Griscelli syndrome type 2
<i>SLC12A6</i>	15	Agenesis of the corpus callosum with peripheral neuropathy
<i>SPRED1</i>	15	Legius syndrome
<i>STRA6</i>	15	Microphthalmia isolated with coloboma 8
<i>UBE3A</i>	15	Angelman syndrome
<i>UBR1</i>	15	Johanson-Blizzard syndrome
<i>ZNF592</i>	15	Spinocerebellar ataxia autosomal recessive 5

<i>ACSF3</i>	16	Combined malonic and methylmalonic aciduria
<i>ALG1</i>	16	Congenital disorder of glycosylation type Ik
<i>ANKRD11</i>	16	KBG syndrome
<i>BBS2</i>	16	Bardet-Biedl syndrome 2
<i>CCDC78</i>	16	Myopathy centronuclear 4
<i>CDH15</i>	16	Mental retardation autosomal dominant 3
<i>COG7</i>	16	Congenital disorder of glycosylation type IIe
<i>COG8</i>	16	Congenital disorder of glycosylation type IIh
<i>CREBBP</i>	16	Rubinstein-Taybi syndrome
<i>FTO</i>	16	Growth retardation developmental delay coarse facies and early death
<i>GCSH</i>	16	Glycine encephalopathy
<i>GPR56</i>	16	Polymicrogyria bilateral frontoparietal
<i>GRIN2A</i>	16	Epilepsy with neurodevelopmental defects
<i>MLYCD</i>	16	Malonyl-CoA decarboxylase deficiency
<i>NDE1</i>	16	Lissencephaly 4 (with microcephaly)
<i>PMM2</i>	16	Congenital disorder of glycosylation type Ia
<i>ROGDI</i>	16	Kohlschutter-Tonz syndrome
<i>RPGRIP1L</i>	16	COACH syndrome
<i>SALL1</i>	16	Townes-Brocks branchiootorenal-like syndrome
<i>SRCAP</i>	16	Floating-Harbor syndrome
<i>TAT</i>	16	Tyrosinemia type II
<i>TBC1D24</i>	16	Epileptic encephalopathy early infantile 16
<i>TMEM231</i>	16	Joubert syndrome 20
<i>TSC2</i>	16	Lymphangiomyomatosis somatic
<i>ACOX1</i>	17	Peroxisomal acyl-CoA oxidase deficiency
<i>ACTG1</i>	17	Baraitser-Winter syndrome 2
<i>ALDH3A2</i>	17	Sjogren-Larsson syndrome
<i>ASPA</i>	17	Canavan disease
<i>COG1</i>	17	Congenital disorder of glycosylation type IIg
<i>EFTUD2</i>	17	Mandibulofacial dysostosis Guion-Almeida type
<i>GFAP</i>	17	Alexander disease
<i>MPDU1</i>	17	Congenital disorder of glycosylation type If

<i>NAGLU</i>	17	Mucopolysaccharidosis type IIIB (Sanfilippo B)
<i>NF1</i>	17	Leukemia juvenile myelomonocytic
<i>PAFAH1B1</i>	17	Lissencephaly 1
<i>PYCR1</i>	17	Cutis laxa autosomal recessive type IIB
<i>RAI1</i>	17	Smith-Magenis syndrome
<i>CCBE1</i>	18	Hennekam lymphangiectasia-lymphedema syndrome
<i>CTDP1</i>	18	Congenital cataracts facial dysmorphism and neuropathy
<i>DYM</i>	18	Dyggve-Melchior-Clausen disease
<i>IER3IP1</i>	18	Microcephaly epilepsy and diabetes syndrome
<i>PIGN</i>	18	Multiple congenital anomalies-hypotonia-seizures syndrome 1
<i>SETBP1</i>	18	Schinzel-Giedion midface retraction syndrome
<i>SMAD4</i>	18	-
<i>TCF4</i>	18	Pitt-Hopkins syndrome
<i>TGIF1</i>	18	-
<i>BCKDHA</i>	19	Maple syrup urine disease type Ia
<i>CC2D1A</i>	19	Mental retardation autosomal recessive 3
<i>DMPK</i>	19	Myotonic dystrophy 1
<i>ERCC2</i>	19	Cerebrooculofacioskeletal syndrome 2
<i>ETHE1</i>	19	Ethylmalonic encephalopathy
<i>FKRP</i>	19	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5
<i>GAMT</i>	19	Cerebral creatine deficiency syndrome 2
<i>MAN2B1</i>	19	Mannosidosis alpha- types I and II
<i>MAP2K2</i>	19	Cardiofaciocutaneous syndrome 4
<i>MCOLN1</i>	19	Mucopolipidosis IV
<i>NDUFA11</i>	19	Mitochondrial complex I deficiency
<i>NDUFS7</i>	19	Leigh syndrome
<i>PEPD</i>	19	Prolidase deficiency
<i>PIK3R2</i>	19	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome
<i>PNKP</i>	19	Epileptic encephalopathy early infantile 10
<i>RNASEH2A</i>	19	Aicardi-Goutieres syndrome 4
<i>SMARCA4</i>	19	Mental retardation autosomal dominant 16

<i>TECR</i>	19	Mental retardation autosomal recessive 14
<i>WDR62</i>	19	Microcephaly 2 primary autosomal recessive with or without cortical malformations
<i>AHCY</i>	20	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
<i>ARFGEF2</i>	20	Periventricular heterotopia with microcephaly
<i>ASXL1</i>	20	Bohring-Opitz syndrome
<i>DNMT3B</i>	20	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
<i>DPM1</i>	20	Congenital disorder of glycosylation type Ie
<i>EPB41L1</i>	20	Mental retardation autosomal dominant 11
<i>GNAS</i>	20	Acromegaly
<i>GSS</i>	20	Glutathione synthetase deficiency
<i>KCNQ2</i>	20	Epileptic encephalopathy early infantile 7
<i>MKKS</i>	20	Bardet-Biedl syndrome 6
<i>PANK2</i>	20	HARP syndrome
<i>PLCB1</i>	20	Epileptic encephalopathy early infantile 12
<i>CBS</i>	21	Homocystinuria B6-responsive and nonresponsive types
<i>DYRK1A</i>	21	Mental retardation autosomal dominant 7
<i>HLCS</i>	21	Holocarboxylase synthetase deficiency
<i>PCNT</i>	21	Microcephalic osteodysplastic primordial dwarfism type II
<i>ACO2</i>	22	Infantile cerebellar-retinal degeneration
<i>ADSL</i>	22	Adenylosuccinase deficiency
<i>ALG12</i>	22	Congenital disorder of glycosylation type Ig
<i>CACNG2</i>	22	Mental retardation autosomal dominant 10
<i>CHKB-</i>	22	-
<i>CPT1B</i>		
<i>CYB5R3</i>	22	Methemoglobinemia type I
<i>EP300</i>	22	Colorectal cancer somatic
<i>LARGE</i>	22	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6
<i>NAGA</i>	22	Kanzaki disease
<i>PEX26</i>	22	Peroxisome biogenesis disorder 7A (Zellweger)
<i>PRODH</i>	22	Hyperprolinemia type I

<i>SCO2</i>	22	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 1
<i>SMARCB1</i>	22	Mental retardation autosomal dominant 15
<i>SNAP29</i>	22	Cerebral dysgenesis neuropathy ichthyosis and palmoplantar keratoderma syndrome
<i>SOX10</i>	22	PCWH syndrome
<i>UPB1</i>	22	Beta-ureidopropionase deficiency
<i>ABCD1</i>	X	Adrenoleukodystrophy
<i>ACSL4</i>	X	Mental retardation X-linked 63
<i>AFF2</i>	X	Mental retardation X-linked FRAXE type
<i>AGTR2</i>	X	Mental retardation X-linked 88
<i>AIFM1</i>	X	Combined oxidative phosphorylation deficiency 6
<i>AP1S2</i>	X	Mental retardation X-linked syndromic Fried type
<i>ARHGEF6</i>	X	Mental retardation X-linked 46
<i>ARHGEF9</i>	X	Epileptic encephalopathy early infantile 8
<i>ARX</i>	X	Epileptic encephalopathy early infantile 1
<i>ATP6AP2</i>	X	Mental retardation X-linked with epilepsy
<i>ATP7A</i>	X	Menkes disease
<i>ATRX</i>	X	Alpha-thalassemia myelodysplasia syndrome somatic
<i>BCOR</i>	X	Microphthalmia syndromic 2
<i>BRWD3</i>	X	Mental retardation X-linked 93
<i>CASK</i>	X	FG syndrome 4
<i>CDKL5</i>	X	Angelman syndrome-like
<i>CUL4B</i>	X	Mental retardation X-linked syndromic 15 (Cabezas type)
<i>DCX</i>	X	Lissencephaly X-linked
<i>DKC1</i>	X	Dyskeratosis congenita X-linked
<i>DLG3</i>	X	Mental retardation X-linked 90
<i>DMD</i>	X	Becker muscular dystrophy
<i>FGD1</i>	X	Aarskog-Scott syndrome
<i>FLNA</i>	X	Cardiac valvular dysplasia X-linked
<i>FMR1</i>	X	Fragile X syndrome
<i>FTSJ1</i>	X	Mental retardation X-linked 9

<i>GDI1</i>	X	Mental retardation X-linked 41
<i>GK</i>	X	Glycerol kinase deficiency
<i>GPC3</i>	X	Simpson-Golabi-Behmel syndrome type 1
<i>GRIA3</i>	X	Mental retardation X-linked 94
<i>HCCS</i>	X	Microphthalmia syndromic 7
<i>HCFC1</i>	X	Mental retardation X-linked 3
<i>HDAC8</i>	X	Cornelia de Lange syndrome 5
<i>HPRT1</i>	X	HPRT-related gout
<i>HSD17B10</i>	X	17-beta-hydroxysteroid dehydrogenase X deficiency
<i>IDS</i>	X	Mucopolysaccharidosis II
<i>IKBKG</i>	X	Ectodermal dysplasia hypohidrotic with immune deficiency
<i>IL1RAPL1</i>	X	Mental retardation X-linked 21/34
<i>IQSEC2</i>	X	Mental retardation X-linked 1
<i>KANSL1</i>	X	Koolen-De Vries syndrome
<i>KDM5C</i>	X	Mental retardation X-linked syndromic Claes-Jensen type
<i>KDM6A</i>	X	Kabuki syndrome 2
<i>KMT2D</i>	X	Kabuki syndrome 1
<i>KRBOX4</i>	X	-
<i>L1CAM</i>	X	Corpus callosum partial agenesis of
<i>LAMP2</i>	X	Danon disease
<i>MAGT1</i>	X	Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia
<i>MAOA</i>	X	Brunner syndrome
<i>MECP2</i>	X	Angelman syndrome
<i>MED12</i>	X	Lujan-Fryns syndrome
<i>MID1</i>	X	Opitz GBBB syndrome type I
<i>MPLKIP</i>	X	Trichothiodystrophy nonphotosensitive 1
<i>NAA10</i>	X	N-terminal acetyltransferase deficiency
<i>NDP</i>	X	Exudative vitreoretinopathy X-linked
<i>NDUFA1</i>	X	Mitochondrial complex I deficiency
<i>NHS</i>	X	Cataract 40 X-linked
<i>NLGN4X</i>	X	-

<i>NSDHL</i>	X	CHILD syndrome
<i>OCRL</i>	X	Dent disease 2
<i>OFD1</i>	X	Joubert syndrome 10
<i>OPHN1</i>	X	Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance
<i>PAK3</i>	X	Mental retardation X-linked 30/47
<i>PCDH19</i>	X	Epileptic encephalopathy early infantile 9
<i>PDHA1</i>	X	Leigh syndrome X-linked
<i>PGK1</i>	X	Phosphoglycerate kinase 1 deficiency
<i>PHF6</i>	X	Borjeson-Forssman-Lehmann syndrome
<i>PHF8</i>	X	Mental retardation syndrome X-linked Siderius type
<i>PLP1</i>	X	Pelizaeus-Merzbacher disease
<i>PORCN</i>	X	Focal dermal hypoplasia
<i>PQBP1</i>	X	Renpenning syndrome
<i>PRPS1</i>	X	Arts syndrome
<i>PTCHD1</i>	X	-
<i>RAB39B</i>	X	Mental retardation X-linked 72
<i>RAB40AL</i>	X	Mental retardation X-linked syndromic Martin-Probst type
<i>RPS6KA3</i>	X	Coffin-Lowry syndrome
<i>SC5D</i>	X	-
<i>SHROOM4</i>	X	Stocco dos Santos X-linked mental retardation syndrome
<i>SLC16A2</i>	X	Allan-Herndon-Dudley syndrome
<i>SLC6A8</i>	X	Cerebral creatine deficiency syndrome 1
<i>SLC9A6</i>	X	Mental retardation X-linked syndromic Christianson type
<i>SMC1A</i>	X	-
<i>SMS</i>	X	Mental retardation X-linked Snyder-Robinson type
<i>SOX3</i>	X	Mental retardation X-linked with isolated growth hormone deficiency
<i>SRPX2</i>	X	Rolandic epilepsy mental retardation and speech dyspraxia
<i>SYN1</i>	X	Epilepsy X-linked with variable learning disabilities and behavior disorders
<i>SYP</i>	X	Mental retardation X-linked 96
<i>TIMM8A</i>	X	Deafness X-linked 1
<i>TSPAN7</i>	X	Mental retardation X-linked 58

<i>UBE2A</i>	X	Mental retardation X-linked syndromic Nascimento-type
<i>UPF3B</i>	X	Mental retardation X-linked syndromic 14
<i>ZDHHC9</i>	X	Mental retardation X-linked syndromic Raymond type
<i>ZNF41</i>	X	Mental retardation X-linked 89
<i>ZNF674</i>	X	Mental retardation X-linked 92
<i>ZNF711</i>	X	Mental retardation X-linked 97
<i>ZNF81</i>	X	Mental retardation X-linked 45
