CHU Liège : Gene Panel - Intellectual Disability v5 (1091 genes)

Gene (HGNC)	OMIM (gene)	OMIM (disease)	OMIM (phenotype)	Fraction of Length Covered Above 30x (%)
AAAS	605378	231550	Achalasia-addisonianism-alacrimia syndrome	99,668
AARS1	601065	616339	Developmental and epileptic encephalopathy 29	99,519
ABAT	137150	613163	GABA-transaminase deficiency	99,682
ABCC9	601439	239850	Hypertrichotic osteochondrodysplasia	99,370
ABCD1	300371	300100	Adrenoleukodystrophy	98,700
ABHD5	604780	275630	Chanarin-Dorfman syndrome	99,068
ACAD9	611103	611126	Mitochondrial complex I deficiency due to ACAD9 deficiency	99,569
ACO2	100850	614559	Infantile cerebellar-retinal degeneration	99,663
ACOX1	609751	264470	Peroxisomal acyl-CoA oxidase deficiency	99,514
ACSF3	614245	614265	Combined malonic and methylmalonic aciduria	99,623
ACSL4	300157	300387	Mental retardation, X-linked 63	91,920
ACTB	102630	243310	Baraitser-Winter syndrome 1	99,483
ACTG1	102560	614583	Baraitser-Winter syndrome 2	99,688
ACTL6B	612458	618468	Developmental and epileptic encephalopathy 76	98,299
ACY1	104620	609924	Aminoacylase 1 deficiency	99,668
ADAMTSL2	612277	231050	Geleophysic dysplasia 1	99,665
ADAR	146920	615010	Aicardi-Goutieres syndrome 6	99,618
ADAT3	615302	615286	Mental retardation, autosomal recessive 36	81,396
ADGRG1	604110	606854	Polymicrogyria, bilateral frontoparietal	99,673
ADK	102750	614300	Hypermethioninemia due to adenosine kinase deficiency	98,599
ADNP	611386	615873	Helsmoortel-van der Aa syndrome	99,661
ADSL	608222	103050	Adenylosuccinase deficiency	99,600
AFF2	300806	309548	Mental retardation, X-linked, FRAXE type	96,053
AFF3	601464	619297	KINSSHIP syndrome	98,120
AFF4	604417	616368	CHOPS syndrome	99,351
AGA	613228	208400	Aspartylglucosaminuria	99,608
AGPAT2	603100	608594	Lipodystrophy, congenital generalized, type 1	89,279
AGPS	603051	600121	Rhizomelic chondrodysplasia punctata, type 3	92,886
AGTPBP1	606830	618276	Neurodegeneration, childhood-onset, with cerebellar atrophy	94,766
AHCY	180960	613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	99,665
AHDC1	615790	615829	Xia-Gibbs syndrome	99,682
AHI1	608894	608629	Joubert syndrome 3	98,968
AIFM1	300169	300816	Combined oxidative phosphorylation deficiency 6	98,330
AIMP1	603605	260600	Leukodystrophy, hypomyelinating, 3	97,431
AKT3	611223	615937	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	98,793
ALDH18A1	138250	219150	Cutis laxa, autosomal recessive, type IIIA	99,661
ALDH3A2	609523	270200	Sjogren-Larsson syndrome	99,283
ALDH4A1	606811	239510	Hyperprolinemia, type II	97,586
ALDH5A1	610045	271980	Succinic semialdehyde dehydrogenase deficiency	86,691
ALDH7A1	107323	266100	Epilepsy, pyridoxine-dependent	99,154
ALG1	605907	608540	Congenital disorder of glycosylation, type Ik	99,237
ALG11	613666	613661	Congenital disorder of glycosylation, type Ip	99,589
ALG12	607144	607143	Congenital disorder of glycosylation, type Ig	99,686
ALG13	300776	300884	Epileptic encephalopathy, early infantile, 36	93,960
ALG2	607905	616228	Myasthenic syndrome, congenital, 14, with tubular aggregates	98,218
ALG3	608750	601110	Congenital disorder of glycosylation, type Id	99,571
ALG6	604566	603147	Congenital disorder of glycosylation, type Ic	98,284
ALG8	608103	608104	Congenital disorder of glycosylation, type Ih	99,171
ALG9	606941	263210	Gillessen-Kaesbach-Nishimura syndrome	97,291
ALMS1	606844	203800	Alstrom syndrome	99,312
ALX1	601527	613456	Frontonasal dysplasia 3	99,588
ALX4	605420	613451	Frontonasal dysplasia 2	98,786
AMER1	300647	300373	Osteopathia striata with cranial sclerosis	99,962
AMPD2	102771	615809	Pontocerebellar hypoplasia, type 9	99,686
AMT	238310	605899	Glycine encephalopathy	99,686
ANK3	600465	615493	Mental retardation, autosomal recessive, 37	99,585
ANKH	605145	123000	Craniometaphyseal dysplasia	99,639
ANKRD11	611192	148050	KBG syndrome	99,648
AP1S1	603531	609313	MEDNIK syndrome	99,440
AP1S2	300629	304340	Mental retardation, X-linked syndromic 5	87,360
AP3B2	602166	617276	Developmental and epileptic encephalopathy 48	97,330
AP4B1	607245	614066	Spastic paraplegia 47, autosomal recessive	99,565
AP4E1	607244	614744	Spastic paraplegia 51, autosomal recessive	98,443
AP4M1	602296	612936	Spastic paraplegia 50, autosomal recessive	99,684
AP4S1	607243	614067	Spastic paraplegia 52, autosomal recessive	99,472
APTX	606350	208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	99,291
ARCN1	600820	617164	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay	99,248
ARFGEF2	605371	608097	Periventricular heterotopia with microcephaly	99,345
ARG1	608313	207800	Argininemia	99,501
ARHGAP31	610911	100300	Adams-Oliver syndrome 1	99,641
ARHGEF2	607560	617523	Neurodevelopmental disorder with midbrain and hindbrain malformations	99,642
ARHGEF9	300429	300607	Epileptic encephalopathy, early infantile, 8	96,726
ARID1A	603024	614607	Coffin-Siris syndrome 2	91,360
ARID1B	614556	135900	Coffin-Siris syndrome 1	90,663
ARID2	609539	617808	Coffis-Siris syndrome 6	99,520
ARL13B	608922	612291	Joubert syndrome 8	98,110
ARL6	608845	600151	Bardet-Biedl syndrome 3	99,068
ARSA	607574	250100	Metachromatic leukodystrophy	99,688
ARSL	300180	302950	Chondrodysplasia punctata, X-linked recessive	92,996
ARX	300382	300419	Mental retardation, X-linked 29 and others	53,864
ASAH1	613468	159950	Spinal muscular atrophy with progressive myoclonic epilepsy	98,326
ASH1L	607999	617796	Mental retardation, autosomal dominant 52	99,562
ASL	608310	207900	Argininosuccinic aciduria	99,678
ASNS	108370	615574	Asparagine synthetase deficiency	99,455
ASPA	608034	271900	Canavan disease	99,125
ASPM	605481	608716	Microcephaly 5, primary, autosomal recessive	98,863
ASS1	603470	215700	Citrullinemia	99,671
ASXL1	612990	605039	Bohring-Opitz syndrome	98,649
ASXL3	615115	615485	Bainbridge-Ropers syndrome	99,408
ATIC	601731	608688	AICA-ribosiduria due to ATIC deficiency	98,266
ATN1	607462	618494	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies	99,114
ATP1A2	182340	602481	Migraine, familial hemiplegic, 2	99,578
ATP1A3	182350	601338	CAPOS syndrome	99,469
ATP5F1A	164360	615228	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4	99,589
ATP6AP2	300556	300423	Mental retardation, X-linked, syndromic, Hedera type	90,146
ATP6V0A2	611716	219200	Cutis laxa, autosomal recessive, type IIA	99,194
ATP6V1A	607027	618012	Developmental and epileptic encephalopathy 93	99,071
ATP7A	300011	309400	Menkes disease	95,512
ATPAF2	608918	604273	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	99,579
ATR	601215	210600	Seckel syndrome 1	99,211
ATRIP	606605	-	Ogi et al., PLoS Genet 2012 : Seckel syndrome	94,377
ATRX	300032	309580	Mental retardation-hypotonic facies syndrome, X-linked	95,087
AUH	600529	250950	3-methylglutaconic aciduria, type I	93,809
AUTS2	607270	615834	Mental retardation, autosomal dominant 26	96,567
B3GALNT2	610194	615181	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	92,556
B3GLCT	610308	261540	Peters-plus syndrome	93,825
B4GALT1	137060	607091	Congenital disorder of glycosylation, type IId	98,272
B4GALT7	604327	130070	Ehlers-Danlos syndrome with short stature and limb anomalies	93,048
B4GAT1	605517	615287	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	99,679
BBIP1	613605	615995	Bardet-Biedl syndrome 18	99,624
BBS1	209901	209900	Bardet-Biedl syndrome 1	99,622
BBS10	610148	615987	Bardet-Biedl syndrome 10	99,484
BBS12	610683	615989	Bardet-Biedl syndrome 12	99,520
BBS2	606151	615981	Bardet-Biedl syndrome 2	99,355
BBS4	600374	615982	Bardet-Biedl syndrome 4	99,511
BBS5	603650	615983	Bardet-Biedl syndrome 5	98,468
BBS7	607590	615984	Bardet-Biedl syndrome 7	99,014
BBS9	615986	615986	Bardet-Biedl syndrome 9	98,795
BCAP31	300398	300475	Deafness, dystonia, and cerebral hypomyelination	92,011
BCKDHA	608348	248600	Maple syrup urine disease, type Ia	99,682
BCKDHB	248611	248600	Maple syrup urine disease, type Ib	98,796
BCKDK	614901	614923	Branched-chain ketoacid dehydrogenase kinase deficiency	99,465
BCL11A	606557	617101	Dias-Logan syndrome	99,609
BCOR	300485	300166	Microphthalmia, syndromic 2	99,025

BCS1L	603647	256000	Leigh syndrome	99,686
BLM	210900	210900	Bloom syndrome	99,077
BOLA3	613183	614299	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	86,569
BRAF	164757	613706	Noonan syndrome 7	94,472
BRAT1	614506	618056	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	99,654
BRF1	604902	616202	Cerebellofaciodental syndrome	97,471
BRPF1	602410	617333	Intellectual developmental disorder with dysmorphic facies and ptosis	99,676
BRSK2	609236	-	Hiatt et al., AJHG 2019	94,939
BRWD3	300553	300659	Mental retardation, X-linked 93	94,284
BSCL2	606158	615924	Encephalopathy, progressive, with or without lipodystrophy	99,677
BTD	609019	253260	Biotinidase deficiency	99,681
BUB1B	602860	257300	Mosaic variegated aneuploidy syndrome 1	99,431
c12orf4	616082	618221	Mental retardation, autosomal recessive 66	99,130
C12ORF57	615640	218340	Temtamy syndrome	99,688
C12orf65	613541	615035	Spastic paraplegia 55, autosomal recessive	98,403
C19ORF12	614297	614298	Neurodegeneration with brain iron accumulation 4	99,688
C2CD3	615944	615948	Orofaciodigital syndrome XIV	99,439
C5ORF42	614571	614615	Joubert syndrome 17	99,024
CA8	114815	613227	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	99,365
CACNA1A	601011	617106	Epileptic encephalopathy, early infantile, 42	97,684
CACNA1C	114205	601005	Timothy syndrome	99,657
CACNA1D	114206	615474	Primary aldosteronism, seizures, and neurologic abnormalities	99,489
CACNA1E	601013	618285	Developmental and epileptic encephalopathy 69	99,614
CACNA2D2	607082	618501	Cerebellar atrophy with seizures and variable developmental delay	94,502
CAD	114010	616457	Developmental and epileptic encephalopathy 50	99,672
CAMK2A	114078	617798	Mental retardation, autosomal dominant 53	99,670
CAMK2B	607707	617799	Mental retardation, autosomal dominant 54	96,977
CANT1	613165	251450	Desbuquois dysplasia 1	99,609
CASK	300172	300749	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	95,518
CAV1	601047	606721	Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome	99,658
CBL	165360	613563	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	99,545
CBS	613381	236200	Homocystinuria, B6-responsive and nonresponsive types	99,686
CC2D1A	610055	608443	Mental retardation, autosomal recessive 3	99,654
CC2D2A	612013	612285	Joubert syndrome 9	99,198
CCBE1	612753	235510	Hennekam lymphangiectasia-lymphedema syndrome 1	98,832
CCDC22	300859	300963	Ritscher-Schinzel syndrome 2	99,900
CCDC39	613798	613807	Ciliary dyskinesia, primary, 14	97,981
CCDC78	614666	614807	Myopathy, centronuclear, 4	99,688
CCDC88C	611204	236600	Hydrocephalus, nonsyndromic, autosomal recessive	99,564
CCND2	123833	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3	99,679
CD96	606037	211750	C syndrome	99,117
CDC42	116952	616737	Takenouchi-Kosaki syndrome	99,394
CDC45	603465	617063	Meier-Gorlin syndrome 7	99,556
CDC6	602627	613805	Meier-Gorlin syndrome 5	99,336
CDH15	114019	612580	Mental retardation, autosomal dominant 3	95,066
CDK10	603464	617694	Al Kaissi syndrome	99,631
CDK13	603309	617360	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	89,010
CDK5RAP2	608201	604804	Microcephaly 3, primary, autosomal recessive	99,471
CDK8	603184	612100	Intellectual developmental disorder with hypotonia and behavioral abnormalities	98,347
CDKL5	300203	300672	Epileptic encephalopathy, early infantile, 2	91,605
CDON	608707	614226	Holoprosencephaly 11	98,960
CDT1	605525	613804	Meier-Gorlin syndrome 4	86,402
CENPJ	609279	608393	Microcephaly 6, primary, autosomal recessive	99,206
CEP104	616690	614615	Joubert syndrome 25	99,525
CEP135	611423	614673	Microcephaly 8, primary, autosomal recessive	97,499
CEP152	613529	614852	Microcephaly 9, primary, autosomal recessive	98,832
CEP290	610142	610188	Joubert syndrome 5	97,702
CEP41	610523	614464	Joubert syndrome 15	99,195
CEP57	607951	614114	Mosaic variegated aneuploidy syndrome 2	98,973
CEP63	614724	614728	Seckel syndrome 6	99,120
CHAMP1	616327	616579	Mental retardation, autosomal dominant 40	99,627
CHAT	118490	254210	Myasthenic syndrome, congenital, 6, presynaptic	97,889
CHD2	602119	615369	Epileptic encephalopathy, childhood-onset	99,277
CHD4	603277	617159	Sifrim-Hitz-Weiss syndrome	99,658
CHD7	608892	214800	CHARGE syndrome	99,585
CHD8	610528	615032	Autism, susceptibility to, 18	99,592
CHKB	612395	602541	Muscular dystrophy, congenital, megaconial type	97,317
CHMP1A	164010	614961	Pontocerebellar hypoplasia, type 8	99,626
CHRNA2	118502	610253	Epilepsy, nocturnal frontal lobe, type 4	99,668
CHRNA4	118504	600513	Epilepsy, nocturnal frontal lobe, 1	97,028
CHRNB2	118507	605375	Epilepsy, nocturnal frontal lobe, 3	99,261
CHRND	100720	616322	Myasthenic syndrome, congenital, 3B, fast-channel	99,685
CISD2	611507	604928	Wolfram syndrome 2	98,682
CLCN2	600570	607628	{Epilepsy, juvenile myoclonic, susceptibility to, 8}	99,685
CLCN4	302910	300114	Mental retardation, X-linked 49/15	99,420
CLN3	607042	204200	Ceroid lipofuscinosis, neuronal, 3	99,687
CLN5	608102	256731	Ceroid lipofuscinosis, neuronal, 5	97,952
CLN6	606725	601780	Ceroid lipofuscinosis, neuronal, 6	89,542
CLN8	607837	600143	Ceroid lipofuscinosis, neuronal, 8	99,688
CLP1	608757	615803	Pontocerebellar hypoplasia, type 10	99,685
CLPB	616254	616271	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	99,616
CLTC	118955	617854	Mental retardation, autosomal dominant 56	99,370
CNKSR2	300724	301008	Mental retardation, X-linked, syndromic, Houge type	93,639
CNNM2	607803	616418	Hypomagnesemia, seizures, and mental retardation	96,206
CNOT3	604910	618672	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies	99,683
CNTNAP1	602346	616286	Lethal congenital contracture syndrome 7	99,653
CNTNAP2	604569	610042	Pitt-Hopkins like syndrome 1	99,356
COA8	616003	220110	Mitochondrial complex IV deficiency, nuclear type 17	97,252
COASY	609855	615643	Neurodegeneration with brain iron accumulation 6	99,684
COG1	606973	611209	Congenital disorder of glycosylation, type IIg	97,762
COG4	606976	613489	Congenital disorder of glycosylation, type IIj	99,615
COG5	606821	613612	Congenital disorder of glycosylation, type IIi	99,097
COG6	606977	614576	Congenital disorder of glycosylation, type III	98,557
COG7	606978	608779	Congenital disorder of glycosylation, type IIe	99,659
COG8	606979	611182	Congenital disorder of glycosylation, type IIh	97,737
COL4A1	120130	175780	Porencephaly 1	98,016
COL4A2	120090	614483	Porencephaly 2	99,620
COL4A3BP	604677	616351	Mental retardation, autosomal dominant 34	99,213
COLEC10	607620	616351	Mental retardation, autosomal dominant 34	99,300
COLEC11	612502	265050	3MC syndrome 2	99,681
COQ2	609825	607426	Coenzyme Q10 deficiency, primary, 1	90,396
COQ4	612898	616276	Coenzyme Q10 deficiency, primary, 7	99,030
COQ6	614647	614650	Coenzyme Q10 deficiency, primary, 6	99,589
COQ7	601683	616733	Coenzyme Q10 deficiency, primary, 8	99,461
COQ8A	606980	612016	Coenzyme Q10 deficiency, primary, 4	99,686
COQ9	612837	614654	Coenzyme Q10 deficiency, primary, 5	98,586
COX10	602125	256000	Leigh syndrome due to mitochondrial COX4 deficiency	99,629
COX15	603646	615119	Mitochondrial complex IV deficiency, nuclear type 6	99,669
CPS1	608307	237300	Carbamoylphosphate synthetase I deficiency	99,520
CRADD	603454	614499	Mental retardation, autosomal recessive 34, with variant lissencephaly	99,635
CRBN	609262	607417	Mental retardation, autosomal recessive 2	99,062
CREBBP	600140	180849	Rubinstein-Taybi syndrome 1	99,645
CRPPA	614631	614643	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	90,229
CSNK2A1	115440	617062	Okur-Chung neurodevelopmental syndrome	99,391
CSPP1	611654	615636	Joubert syndrome 21	98,895
CSTB	601145	254800	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	91,900
CTC1	613129	612199	Cerebroretinal microangiopathy with calcifications and cysts	99,598
CTCF	604167	615502	Mental retardation, autosomal dominant 21	99,599
CTNNB1	116806	615075	Mental retardation, autosomal dominant 19	99,651
CTSA	613111	256540	Galactosialidosis	99,583
CTSD	116840	610127	Ceroid lipofuscinosis, neuronal, 10	96,685
CUL3	603136	619239	Neurodevelopmental disorder with or without autism or seizures	99,122
CUL4B	300304	300354	Mental retardation, X-linked, syndromic 15 (Cabezas type)	87,698
CWF19L1	616120	616127	Spinocerebellar ataxia, autosomal recessive 17	99,503
CYB5R3	613213	250800	Methemoglobinemia, type I	95,256
CYFIP2	606323	618468	Developmental and epileptic encephalopathy 65	99,634
CYP27A1	606530	213700	Cerebrotendinous xanthomatosis	97,278
CYP2U1	610670	615030	Spastic paraplegia 56, autosomal recessive	90,599
D2HGDH	609186	600721	D-2-hydroxyglutaric aciduria	97,812
DAG1	128239	616538	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9	99,688

DARS1	603084	615281	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	98,569
DARS2	610956	611105	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	99,194
DBT	248610	248600	Maple syrup urine disease, type II	98,714
DCAF17	612515	241080	Woodhouse-Sakati syndrome	93,250
DCC	120470	157600	Mirror movements 1 and/or agenesis of the corpus callosum	99,550
DCPS	610534	616459	Al-Raqad syndrome	99,687
DCX	300121	300067	Lissencephaly, X-linked	97,662
DDB2	600811	278740	Xeroderma pigmentosum, group E, DDB-negative subtype	99,687
DDC	107930	608643	Aromatic L-amino acid decarboxylase deficiency	99,487
DDHD2	615003	615033	Spastic paraplegia 54, autosomal recessive	98,212
DDX11	601150	613398	Warsaw breakage syndrome	99,652
DDX3X	300160	300958	Mental retardation, X-linked 102	97,259
DEAF1	602635	615828	Mental retardation, autosomal dominant 24	86,514
DEGS1	615843	618404	Leukodystrophy, hypomyelinating, 18	97,990
DEPDC5	614191	604364	Epilepsy, familial focal, with variable foci 1	99,497
DHCR24	606418	602398	Desmosterolosis	99,238
DHCR7	602858	270400	Smith-Lemli-Opitz syndrome	99,678
DHDDS	608172	617836	Developmental delay and seizures with or without movement abnormalities	99,503
DHFR	126060	613839	Megaloblastic anemia due to dihydrofolate reductase deficiency	98,428
DHX30	616423	617804	Neurodevelopmental disorder with severe motor impairment and absent language	99,550
DIAPH1	602121	616632	Seizures, cortical blindness, microcephaly syndrome	96,283
DKC1	300126	305000	Dyskeratosis congenita, X-linked	93,593
DLD	238331	246900	Dihydrolipoamide dehydrogenase deficiency	99,071
DLG3	300189	300850	Mental retardation, X-linked 90	98,723
DLL4	605185	616589	Adams-Oliver syndrome 6	99,648
DNAJC19	608977	610198	3-methylglutaconic aciduria, type V	97,290
DNM1	602377	616346	Epileptic encephalopathy, early infantile, 31	94,954
DNMT3A	602769	615879	Tatton-Brown-Rahman syndrome	99,685
DNMT3B	602900	242860	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	99,686
DOCK6	614194	614219	Adams-Oliver syndrome 2	98,871
DOCK7	615730	615859	Developmental and epileptic encephalopathy 23	98,235
DOCK8	611432	243700	Hyper-IgE recurrent infection syndrome, autosomal recessive	99,383
DOLK	610746	610768	Congenital disorder of glycosylation, type Im	99,684
DPAGT1	191350	608093	Congenital disorder of glycosylation, type Ij	99,684
DPH1	603527	616901	Developmental delay with short stature, dysmorphic features, and sparse hair	99,667
DPM1	603503	608799	Congenital disorder of glycosylation, type Ie	99,282
DPM2	603564	615042	Congenital disorder of glycosylation, type Iu	99,688
DPP6	126141	616311	Mental retardation, autosomal dominant 33	99,567
DSCAM	602523	-	Wang et al., Nat Commun 2016 : Autisme	99,500
DYNC1H1	600112	614563	Mental retardation, autosomal dominant 13	99,467
DYRK1A	600855	614104	Mental retardation, autosomal dominant 7	99,560
EARS2	612799	614924	Combined oxidative phosphorylation deficiency 12	99,651
EBF3	607407	617330	Hypotonia, ataxia, and delayed development syndrome	98,428
EBP	300205	300960	MEND syndrome	99,874
ECHS1	602292	616277	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	98,673
EDC3	609842	616460	Mental retardation, autosomal recessive 50	99,676
EEF1A2	602959	616393	Mental retardation, autosomal dominant 38	92,159
EFNB1	300035	304110	Craniofrontonasal dysplasia	99,590
EFTUD2	603892	610536	Mandibulofacial dysostosis, Guion-Almeida type	99,426
EHMT1	607001	610253	Kleefstra syndrome	97,929
EIF2AK3	604032	226980	Wolcott-Rallison syndrome	92,143
EIF2B1	606686	603896	Leukoencephalopathy with vanishing white matter	99,599
EIF2B2	606454	603897	Leukoencephalopathy with vanishing white matter	98,066
EIF2B3	606273	603898	Leukoencephalopathy with vanishing white matter	99,569
EIF2B4	606687	603899	Leukoencephalopathy with vanishing white matter	99,634
EIF2B5	603945	603900	Leukoencephalopathy with vanishing white matter	99,614
EIF2S3	300161	300987	Mental retardation, X-linked, syndromic, Borck type	98,000
EIF3F	603914	618295	Mental retardation, autosomal recessive 67	99,633
ELAC2	605367	615440	Combined oxidative phosphorylation deficiency 17	98,763
ELOVL4	605512	614457	Ichthyosis, spastic quadriplegia, and mental retardation	99,430
ELP1	603722	223900	Dysautonomia, familial	99,529
ELP2	616054	617270	Mental retardation, autosomal recessive 58	98,990
EML1	602033	600348	Band heterotopia	97,331
EMX2	600035	269160	Schizencephaly	99,370
ENTPD1	601752	615683	Spastic paraplegia 64, autosomal recessive	99,173
EOGT	614789	615297	Adams-Oliver syndrome 4	99,304
EP300	602700	613684	Rubinstein-Taybi syndrome 2	99,590
EPB41L1	602879	614257	Mental retardation, autosomal dominant 11	99,686
EPG5	615068	242840	Vici syndrome	99,484
EPM2A	607566	254780	Epilepsy, progressive myoclonic 2A (Lafora)	80,285
ERCC2	126340	610756	Cerebrooculofacioskeletal syndrome 2	99,664
ERCC3	133510	610651	Xeroderma pigmentosum, group B	99,683
ERCC4	133520	278760	Xeroderma pigmentosum, type F/Cockayne syndrome	99,434
ERCC5	133530	278780	Xeroderma pigmentosum, group G/Cockayne syndrome	99,476
ERCC6	609413	214150	Cerebrooculofacioskeletal syndrome 1	99,318
ERCC8	609412	216400	Cockayne syndrome, type A	99,283
ERLIN2	611605	611225	Spastic paraplegia 18, autosomal recessive	99,335
ESCO2	609353	268300	Roberts syndrome	97,834
ETHE1	608451	602473	Ethylmalonic encephalopathy	99,619
EXOSC3	606489	614678	Pontocerebellar hypoplasia, type 1B	99,526
EZH2	601573	277590	Weaver syndrome	99,407
EZR	123900	-	Riecken et al., Hum Mut 2015 : ID	99,645
FA2H	611026	612319	Spastic paraplegia 35, autosomal recessive	87,408
FAM126A	610531	610532	Leukodystrophy, hypomyelinating, 5	99,109
FARS2	611592	614946	Combined oxidative phosphorylation deficiency 14	99,595
FASTKD2	612322	618855	Combined oxidative phosphorylation deficiency 44	99,315
FAT4	612411	615546	Van Maldergem syndrome 2	99,617
FBXL4	605654	615471	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	99,498
FBXO11	607871	618089	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities	94,198
FBXO31	609102	615979	Mental retardation, autosomal recessive 45	88,448
FGD1	300546	305400	Mental retardation, X-linked syndromic 16	97,166
FGF12	601513	617166	Developmental and epileptic encephalopathy 47	99,461
FGF14	601515	609307	Spinocerebellar ataxia 27	98,854
FGFR1	136350	147950	Hypogonadotropic hypogonadism 2 with or without anosmia	99,687
FGFR2	176943	101400	Saethre-Chotzen syndrome	99,674
FGFR3	134934	612247	Crouzon syndrome with acanthosis nigricans	96,608
FH	136850	606812	Fumarase deficiency	99,314
FKRP	606596	606612	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5	85,031
FKTN	607440	613152	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4	99,076
FLNA	300017	300049	Heterotopia, periventricular	99,553
FMN2	606373	616193	Mental retardation, autosomal recessive 47	97,538
FMR1	309550	300623	Fragile X tremor/ataxia syndrome	94,957
FOLR1	136430	613068	Neurodegeneration due to cerebral folate transport deficiency	99,688
FOXG1	164874	613454	Rett syndrome, congenital variant	84,195
FOXP1	605515	613670	Mental retardation with language impairment and with or without autistic features	99,569
FOXP2	605317	602081	Speech-language disorder-1	99,415
FOXRED1	613622	256000	Leigh syndrome due to mitochondrial complex I deficiency	99,633
FRAS1	607830	219000	Fraser syndrome 1	99,576
FREM2	608945	617666	Fraser syndrome 2	99,500
FRMPD4	300838	300983	Mental retardation, X-linked 104	98,022
FTCD	606806	229100	Glutamate formiminotransferase deficiency	95,444
FTL	134790	606159	Neurodegeneration with brain iron accumulation 3	99,550
FTO	610966	612938	Growth retardation, developmental delay, facial dysmorphism	99,532
FTSJ1	300499	309549	Mental retardation, X-linked 9/44	99,827
FUCA1	612280	230000	Fucosidosis	96,756
GABBR2	607340	617904	Developmental and epileptic encephalopathy 59	95,408
GABRA1	137160	615744	Developmental and epileptic encephalopathy 19	99,632
GABRA2	137140	618557	Developmental and epileptic encephalopathy 78	99,309
GABRA3	305660	-	Niturad et al., Brain 2017 : range of epileptic seizure types, a varying degree of intellectual disability and developmental delay, sometimes with dysmorphic features or nystagmus.	92,805
GABRA5	137142	618559	Developmental and epileptic encephalopathy 79	99,645
GABRB2	600232	617829	Developmental and epileptic encephalopathy 92	99,556
GABRB3	137192	617113	Epileptic encephalopathy, early infantile, 43	96,677
GABRG2	137164	611277	Epilepsy, generalized, with febrile seizures plus, type 3	99,571
GAD1	605363	619124	Developmental and epileptic encephalopathy 89	99,664
GALC	606890	245200	Krabbe disease	94,581
GALT	606999	230400	Galactosemia	99,671
GAMT	601240	612736	Cerebral creatine deficiency syndrome 2	90,752
GAN	605379	256850	Giant axonal neuropathy-1	98,717
GATAD2B	614998	615074	Mental retardation, autosomal dominant 18	99,686

GATM	602360	612718	Cerebral creatine deficiency syndrome 3	96,388
GBA2	609471	614409	Spastic paraplegia 46, autosomal recessive	99,686
GBE1	607839	232500	Glycogen storage disease IV	98,823
GCDH	608801	231670	Glutaricaciduria, type I	99,687
GCH1	600225	128230	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	87,483
GCSH	238330	605899	Glycine encephalopathy	81,534
GDI1	300104	300849	Mental retardation, X-linked 41	97,599
GFAP	137780	203450	Alexander disease	99,685
GFM1	606639	609060	Combined oxidative phosphorylation deficiency 1	98,618
GJC2	608803	608804	Leukodystrophy, hypomyelinating, 2	73,702
GLB1	611458	230500	GM1-gangliosidosis, type I	99,577
GLDC	238300	605899	Glycine encephalopathy	96,244
GLI2	165230	610829	Holoprosencephaly 9	92,829
GLI3	165240	175700	Greig cephalopolysyndactyly syndrome	99,327
GLRA1	138491	149600	Hyperekplexia 1	99,523
GLRB	138492	614619	Hyperekplexia 2	98,634
GLUL	138290	610015	Glutamine deficiency, congenital	99,678
GLYCTK	610516	220120	D-glyceric aciduria	99,688
GM2A	613109	272750	GM2-gangliosidosis, AB variant	99,678
GMPPA	615495	615510	Alacrima, achalasia, and mental retardation syndrome	99,688
GMPPB	615320	615351	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14	99,644
GNAI3	139370	602483	Auriculocondylar syndrome 1	99,076
GNAO1	139311	615473	Epileptic encephalopathy, early infantile, 17	99,659
GNAS	139320	103580	Pseudohypoparathyroidism Ia	97,261
GNB1	139380	616973	Mental retardation, autosomal dominant 42	99,429
GNPAT	602744	222765	Rhizomelic chondrodysplasia punctata, type 2	97,788
GNPTAB	607840	252500	Mucolipidosis II alpha/beta	99,517
GNPTG	607838	252605	Mucolipidosis III gamma	92,848
GNS	607664	252940	Mucopolysaccharidosis type IIID	99,502
GOSR2	604027	614018	Epilepsy, progressive myoclonic 6	99,669
GPC3	300037	312870	Simpson-Golabi-Behmel syndrome, type 1	94,597
GPHN	603930	615501	Molybdenum cofactor deficiency C	99,589
GPSM2	609245	604213	Chudley-McCullough syndrome	99,202
GPT2	138210	616281	Mental retardation, autosomal recessive 49	92,577
GRIA3	305915	300699	Mental retardation, X-linked 94	91,651
GRIA4	138246	617864	Neurodevelopmental disorder with or without seizures and gait abnormalities	99,149
GRID2	602368	616204	Spinocerebellar ataxia, autosomal recessive 18	99,524
GRIK2	138244	611092	Mental retardation, autosomal recessive, 6	99,225
GRIN1	138249	614254	Mental retardation, autosomal dominant 8	99,469
GRIN2A	138253	245570	Epilepsy, focal, with speech disorder and with or without mental retardation	99,682
GRIN2B	138252	613970	Mental retardation, autosomal dominant 6	99,671
GRIN2D	602717	617162	Developmental and epileptic encephalopathy 46	62,309
GRIP1	604597	617667	Fraser syndrome 3	99,651
GRM1	604473	617691	Spinocerebellar ataxia 44	99,548
GSS	601002	266130	Glutathione synthetase deficiency	99,595
GTF2E2	189964	616943	Trichothiodystrophy 6, nonphotosensitive	98,910
GTF2H5	608780	616395	Trichothiodystrophy 3, photosensitive	99,609
GTPBP2	607434	617988	Jaberi-Elahi syndrome	97,964
GTPBP3	608536	616198	Combined oxidative phosphorylation deficiency 23	96,840
GUSB	611499	253220	Mucopolysaccharidosis VII	98,515
HACE1	610876	616756	Spastic paraplegia and psychomotor retardation with or without seizures	97,131
HAX1	605998	610738	Neutropenia, severe congenital 3, autosomal recessive	99,680
HCFC1	300019	616756	Spastic paraplegia and psychomotor retardation with or without seizures	99,847
HCN1	602780	615871	Epileptic encephalopathy, early infantile, 24	92,793
HDAC4	605314	-	Fisch et al., AJMG 2016 : ID	99,676
HDAC8	300269	300882	Cornelia de Lange syndrome 5	99,211
HECW2	617245	617268	Neurodevelopmental disorder with hypotonia, seizures, and absent language	99,538
HEPACAM	611642	613926	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation	86,263
HERC1	605109	617011	Macrocephaly, dysmorphic facies, and psychomotor retardation	99,543
HERC2	605837	615516	Mental retardation, autosomal recessive 38	99,667
HESX1	601802	182230	Growth hormone deficiency with pituitary anomalies	98,652
HEXA	606869	272800	Tay-Sachs disease	99,673
HEXB	606873	268800	Sandhoff disease, infantile, juvenile, and adult forms	96,791
HGSNAT	610453	252930	Mucopolysaccharidosis type IIIC (Sanfilippo C)	93,515
HIBCH	610690	250620	3-hydroxyisobutryl-CoA hydrolase deficiency	99,303
HIVEP2	143054	616977	Mental retardation, autosomal dominant 43	99,666
HLCS	609018	253270	Holocarboxylase synthetase deficiency	99,620
HNMT	605238	616739	Mental retardation, autosomal recessive 51	99,399
HNRNPH2	300610	300986	Mental retardation, X-linked, syndromic, Bain type	99,854
HNRNPK	600712	616580	Au-Kline syndrome	99,213
HNRNPU	602869	617391	Epileptic encephalopathy, early infantile, 54	95,271
HOXA1	142955	601536	Athabaskan brainstem dysgenesis syndrome	99,657
HPD	609695	276710	Tyrosinemia, type III	99,666
HPRT1	308000	300322	Lesch-Nyhan syndrome	82,375
HRAS	190020	218040	Costello syndrome	99,688
HSD17B10	300256	300438	HSD10 mitochondrial disease	99,971
HSPD1	118190	612233	Leukodystrophy, hypomyelinating, 4	99,436
HSPG2	142461	224410	Dyssegmental dysplasia, Silverman-Handmaker type	99,070
HUWE1	300697	300706	Mental retardation, X-linked syndromic, Turner type	97,923
IARS1	600709	617093	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	99,316
IARS2	612801	616007	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	98,936
IDS	300823	309900	Mucopolysaccharidosis II	97,839
IDUA	252800	607015	Mucopolysaccharidosis Ih/s	74,816
IER3IP1	609382	614231	Microcephaly, epilepsy, and diabetes syndrome	99,061
IFIH1	606951	615846	Aicardi-Goutieres syndrome 7	99,116
IFT172	607386	615630	Short-rib thoracic dysplasia 10 with or without polydactyly	99,632
IFT27	615870	615996	Bardet-Biedl syndrome 19	99,688
IFT57	606621	617927	?Orofaciodigital syndrome XVIII	98,894
IGF1	147440	608747	Growth retardation with deafness and mental retardation due to IGF1 deficiency	99,625
IL1RAPL1	300206	300143	Mental retardation, X-linked 21/34	96,043
INPP5E	613037	213300	Joubert syndrome 1	93,816
INTU	610621	617926	?Orofaciodigital syndrome XVII	98,803
IQSEC2	300522	309530	Mental retardation, X-linked 1/78	93,756
IRX5	606195	611174	Hamamy syndrome	88,234
ITGA7	600536	613204	Muscular dystrophy, congenital, due to ITGA7 deficiency	99,670
ITPR1	147265	117360	Spinocerebellar ataxia 29, congenital nonprogressive	99,417
JAM3	606871	613730	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	99,278
KANSL1	612452	610443	Koolen-De Vries syndrome	99,654
KAT6A	601408	616268	Mental retardation, autosomal dominant 32	99,646
KAT6B	605880	606170	Genitopatellar syndrome	99,495
KCNA1	176260	160120	Episodic ataxia/myokymia syndrome	99,675
KCNA2	176262	616366	Developmental and epileptic encephalopathy 32	99,688
KCNB1	600397	616056	Developmental and epileptic encephalopathy 26	98,981
KCNC1	176258	616187	Epilepsy, progressive myoclonic 7	98,881
KCNC3	176264	605259	Spinocerebellar ataxia 13	75,162
KCNH1	603305	611816	Temple-Baraitser syndrome	99,156
KCNJ10	602208	612780	SESAME syndrome	99,678
KCNJ11	600937	606176	Diabetes, permanent neonatal, with or without neurologic features	99,688
KCNK9	605874	612292	Birk-Barel mental retardation dysmorphism syndrome	99,681
KCNMA1	600150	609446	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy	99,481
KCNQ2	602235	613720	Epileptic encephalopathy, early infantile, 7	98,281
KCNQ3	602232	121201	Seizures, benign neonatal, 2	94,939
KCNQ5	607357	617601	Mental retardation, autosomal dominant 46	94,217
KCNT1	608167	614959	Epileptic encephalopathy, early infantile, 14	98,370
KCNT2	610044	617771	Developmental and epileptic encephalopathy 57	98,877
KCTD7	611725	611726	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	96,545
KDM1A	609132	616728	Cleft palate, psychomotor retardation, and distinctive facial features	93,390
KDM5C	314690	300534	Mental retardation, X-linked, syndromic, Claes-Jensen type	99,902
KDM6A	300128	300867	Kabuki syndrome 2	95,185
KIAA0586	610178	616490	Joubert syndrome 23	93,835
KIAA0753	617112	617127	Orofaciodigital syndrome XV	99,355
KIAA2022	300524	300912	Mental retardation, X-linked 98	99,714
KIF11	148760	152950	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation	98,782
KIF14	611279	616258	Meckel syndrome 12	98,732
KIF1A	601255	614255	Mental retardation, autosomal dominant 9	99,647
KIF2A	602591	615411	Cortical dysplasia, complex, with other brain malformations 3	98,174
KIF4A	300521	300923	Mental retardation, X-linked 100	96,590

KIF5A	602821	617235	Myoclonus, intractable, neonatal	99,421
KIF5C	604593	615282	Cortical dysplasia, complex, with other brain malformations 2	99,366
KIF7	611254	200990	Joubert syndrome 12	93,224
KIFBP	609367	609460	Goldberg-Shprintzen megacolon syndrome	99,484
KIRREL3	607761	612581	Mental retardation, autosomal dominant 4	99,659
KLC2	611729	609541	Spastic paraplegia, optic atrophy, and neuropathy	99,687
KMT2A	159555	605130	Wiedemann-Steiner syndrome	97,548
KMT2C	606833	617768	Kleefstra syndrome 2	99,461
KMT2D	602113	147920	Kabuki syndrome 1	99,675
KMT2E	608444	618512	O'Donnell-Luria-Rodan syndrome	99,012
KMT5B	610881	617788	Mental retardation, autosomal dominant 51	99,497
KNL1	609173	604321	Microcephaly 4, primary, autosomal recessive	97,122
KPTN	615620	615637	Mental retardation, autosomal recessive 41	99,322
KRAS	190070	609942	Noonan syndrome 3	99,348
L1CAM	308840	303350	CRASH syndrome	99,659
L2HGDH	609584	236792	L-2-hydroxyglutaric aciduria	98,750
LAMA1	150320	615960	Poretti-Boltshauser syndrome	99,356
LAMA2	156225	607855	Muscular dystrophy, congenital, due to partial LAMA2 deficiency	99,479
LAMB1	150240	615191	Lissencephaly 5	99,505
LAMC3	604349	614115	Cortical malformations, occipital	96,238
LAMP2	309060	300257	Danon disease	93,527
LARGE1	603590	608840	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	99,650
LARP7	612026	615071	Alazami syndrome	97,440
LAS1L	300964	309585	Wilson-Turner syndrome	99,790
LIAS	607031	614462	Hyperglycinemia, lactic acidosis, and seizures	99,048
LIG4	601837	606593	LIG4 syndrome	99,554
LINS1	610350	614340	Mental retardation, autosomal recessive 27	99,084
LIPT1	610284	616299	Lipoyltransferase 1 deficiency	98,695
LMBRD1	612625	277380	Methylmalonic aciduria and homocystinuria, cblF type	96,873
LMNA	150330	616516	Emery-Dreifuss muscular dystrophy 3, AR	99,301
LRP2	600073	222448	Donnai-Barrow syndrome	99,435
LRPPRC	607544	220111	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	97,951
LZTFL1	606568	615994	Bardet-Biedl syndrome 17	98,793
LZTR1	600574	616564	Noonan syndrome 10	99,525
MAF	177075	601088	Ayme-Gripp syndrome	73,068
MAGEL2	605283	615547	Schaaf-Yang syndrome	98,062
MAN1B1	604346	614202	Mental retardation, autosomal recessive 15	98,641
MAN2B1	609458	248500	Mannosidosis, alpha-, types I and II	99,413
MANBA	609489	248510	Mannosidosis, beta	99,102
MAOA	309850	300615	Brunner syndrome	90,158
MAP2K1	176872	615279	Cardiofaciocutaneous syndrome 3	99,627
MAP2K2	601263	615280	Cardiofaciocutaneous syndrome 4	99,094
MASP1	600521	257920	3MC syndrome 1	99,614
MAT1A	610550	250850	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	99,655
MBD5	611472	156200	Mental retardation, autosomal dominant 1	99,639
MBOAT7	606048	617188	Mental retardation, autosomal recessive 57	99,373
MBTPS2	300294	308205	IFAP syndrome with or without BRESHECK syndrome	96,335
MCCC1	609010	210200	3-Methylcrotonyl-CoA carboxylase 1 deficiency	99,500
MCOLN1	605248	252650	Mucolipidosis IV	97,195
MCPH1	607117	251200	Microcephaly 1, primary, autosomal recessive	98,777
MECP2	300005	312750	Rett syndrome	97,884
MED12	300188	305450	Opitz-Kaveggia syndrome	99,834
MED13	603808	618009	Intellectual developmental disorder 61	99,021
MED13L	608771	616789	Mental retardation and distinctive facial features with or without cardiac defects	99,573
MED17	603810	613668	Microcephaly, postnatal progressive, with seizures and brain atrophy	98,898
MED23	605042	614249	Mental retardation, autosomal recessive 18	99,122
MEF2C	600662	613443	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	99,545
MEIS2	601740	600987	Cleft palate, cardiac defects, and mental retardation	99,656
METTL23	615262	615942	Mental retardation, autosomal recessive 44	99,621
MFF	614785	617086	Encephalopathy due to defective mitochondrial and peroxisomal fission 2	99,199
MFSD2A	614397	616486	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	99,476
MFSD8	611124	610951	Ceroid lipofuscinosis, neuronal, 7	99,384
MGAT2	602616	212066	Congenital disorder of glycosylation, type IIa	99,548
MICU1	605084	615673	Myopathy with extrapyramidal signs	99,541
MID1	300552	300000	Opitz GBBB syndrome, type I	98,566
MKKS	604896	605231	Bardet-Biedl syndrome 6	99,583
MKS1	609883	615990	Bardet-Biedl syndrome 13	99,532
MLC1	605908	604004	Megalencephalic leukoencephalopathy with subcortical cysts	99,616
MLYCD	606761	248360	Malonyl-CoA decarboxylase deficiency	78,514
MMACHC	609831	277400	Methylmalonic aciduria and homocystinuria, cblC type	99,685
MMADHC	611935	277410	Homocystinuria and Methylmalonic Aciduria	99,148
MOCS1	603707	252150	Molybdenum cofactor deficiency A	99,678
MOCS2	603708	252160	Molybdenum cofactor deficiency B	99,421
MOGS	601336	606056	Congenital disorder of glycosylation, type IIb	95,383
MORC2	616661	619090	Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy	99,679
MPDU1	604041	609180	Congenital disorder of glycosylation, type If	99,687
MPDZ	603785	615219	Hydrocephalus, nonsyndromic, autosomal recessive 2	99,290
MPLKIP	609188	234050	Trichothiodystrophy 4, nonphotosensitive	99,636
MRAS	608435	618499	Noonan syndrome 11	99,659
MSL3	300609	301032	Basilicata-Akhtar syndrome	96,748
MTFMT	611766	614947	Combined oxidative phosphorylation deficiency 15	96,475
MTHFR	607093	236250	Homocystinuria due to MTHFR deficiency	99,542
MTM1	300415	310400	Myotubular myopathy, X-linked	94,570
MTOR	601231	616638	Smith-Kingsmore syndrome	99,598
MTR	156570	250940	Homocystinuria-megaloblastic anemia, cblG complementation type	99,544
MTRR	602568	236270	Homocystinuria-megaloblastic anemia, cbl E type	99,357
MVK	251170	610377	Mevalonic aciduria	99,686
MYCN	164840	164280	Feingold syndrome 1	80,770
MYO5A	160777	214450	Griscelli syndrome, type 1	98,717
MYT1L	613084	616521	Mental retardation, autosomal dominant 39	99,563
NAA10	300013	300855	Ogden syndrome	94,711
NAA15	608000	617787	Mental retardation, autosomal dominant 50	97,593
NACC1	610672	617393	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination	99,617
NAGA	104170	609241	Schindler disease, type I	99,676
NAGLU	609701	252920	Mucopolysaccharidosis type IIIB (Sanfilippo B)	83,856
NALCN	611549	616266	Congenital contractures of the limbs and face, hypotonia, and developmental delay	99,405
NANS	605202	610442	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	99,661
NAPB	611270	-	Conroy et al., Clin Genet 2016 : early-onset epileptic encephalopathy	99,425
NARS1	108410	619091	Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive	99,110
NARS2	612803	616239	Combined oxidative phosphorylation deficiency 24	99,066
NBN	602667	251260	Nijmegen breakage syndrome	98,843
NCDN	608458	-	Fatima et al., AJHG 2021	99,681
NDE1	609449	614019	Lissencephaly 4 (with microcephaly)	99,686
NDP	300658	310600	Norrie disease	99,609
NDST1	600853	616116	Mental retardation, autosomal recessive 46	99,668
NDUFA1	300078	252010	Mitochondrial complex I deficiency	97,308
NDUFA11	612638	252010	Mitochondrial complex I deficiency	99,688
NDUFA12	614530	256000	Leigh syndrome due to mitochondrial complex 1 deficiency	99,649
NDUFA2	602137	256000	Leigh syndrome due to mitochondrial complex I deficiency	99,688
NDUFA9	603834	256000	Leigh syndrome due to mitochondrial complex I deficiency	99,233
NDUFAF2	609653	618233	Mitochondrial complex I deficiency, nuclear type 10	98,730
NDUFAF3	612911	618240	Mitochondrial complex I deficiency, nuclear type 18	99,383
NDUFAF4	611776	618237	Mitochondrial complex I deficiency, nuclear type 15	98,282
NDUFAF5	612360	618238	Mitochondrial complex I deficiency, nuclear type 16	99,094
NDUFAF6	612392	256000	Leigh syndrome due to mitochondrial complex I deficiency	84,034
NDUFB3	603839	618246	Mitochondrial complex I deficiency, nuclear type 25	99,251
NDUFS1	157655	618226	Mitochondrial complex I deficiency, nuclear type 5	99,144
NDUFS2	602985	252010	Mitochondrial complex I deficiency	99,245
NDUFS3	603846	256000	Leigh syndrome due to mitochondrial complex I deficiency	99,673
NDUFS4	602694	252010	Mitochondrial complex I deficiency	98,800
NDUFS6	603848	618232	Mitochondrial complex I deficiency, nuclear type 9	98,031
NDUFS7	601825	256000	Leigh syndrome	99,554
NDUFS8	602141	618222	Mitochondrial complex I deficiency, nuclear type 2	99,669
NDUFV1	161015	618225	Mitochondrial complex I deficiency, nuclear type 4	99,474
NDUFV2	600532	618229	Mitochondrial complex I deficiency, nuclear type 7	97,345
NECAP1	611623	615833	Developmental and epileptic encephalopathy 21	99,535
NEDD4L	606384	617201	Periventricular nodular heterotopia 7	99,066

NEU1	608272	256550	Sialidosis	99,688
NFIA	600727	613735	Brain malformations and urinary tract defects	99,558
NFIX	164005	614753	Sotos syndrome 2	99,638
NGF	162030	608654	Neuropathy, hereditary sensory and autonomic, type V	99,658
NGLY1	610661	615273	Congenital disorder of deglycosylation	99,160
NHEJ1	611290	611291	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	99,674
NHLRC1	608072	254780	Epilepsy, progressive myoclonic 2B (Lafora)	97,843
NHP2	606470	613987	Dyskeratosis congenita, autosomal recessive 2	99,634
NHS	300457	302350	Nance-Horan syndrome	90,557
NIN	608684	614851	Seckel syndrome 7	99,371
NIPBL	608667	122470	Cornelia de Lange syndrome 1	98,718
NKAP	300766	301039	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type	94,900
NKX2-1	600635	610978	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	86,337
NONO	300084	300967	Mental retardation, X-linked, syndromic 34	98,511
NOTCH1	190198	616028	Adams-Oliver syndrome 5	98,524
NPC1	607623	257220	Niemann-Pick disease, type C1	98,964
NPC2	601015	607625	Niemann-pick disease, type C2	99,672
NPHP1	607100	609583	Joubert syndrome 4	99,334
NPHP3	608002	267010	Meckel syndrome 7	97,537
NPRL2	607072	617116	Epilepsy, familial focal, with variable foci 2	99,688
NPRL3	600928	617118	Epilepsy, familial focal, with variable foci 3	99,672
NR2F1	132890	615722	Bosch-Boonstra-Schaaf optic atrophy syndrome	85,529
NR5A1	184757	612964	Adrenocortical insufficiency	99,350
NRAS	164790	613224	Noonan syndrome 6	99,599
NRXN1	600565	614325	Pitt-Hopkins-like syndrome 2	99,551
NSD1	606681	117550	Sotos syndrome 1	99,495
NSDHL	300275	308050	CHILD syndrome	99,472
NSUN2	610916	611091	Mental retardation, autosomal recessive 5	96,483
NTNG2	618689	618718	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia	88,978
NUBPL	613621	618242	Mitochondrial complex I deficiency, nuclear type 21	98,946
OCLN	602876	251290	Pseudo-TORCH syndrome 1	99,503
OCRL	300535	309000	Lowe syndrome	93,920
OFD1	300170	311200	Orofaciodigital syndrome I	92,424
OGT	300255	300997	Mental retardation, X-linked 106	96,387
OPHN1	300127	300486	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	91,854
ORC1	601902	224690	Meier-Gorlin syndrome 1	99,516
ORC4	603056	613800	Meier-Gorlin syndrome 2	98,665
ORC6	607213	613803	Meier-Gorlin syndrome 3	98,116
OSGEP	610107	617729	Galloway-Mowat syndrome 3	99,648
OTC	300461	311250	Ornithine transcarbamylase deficiency	92,394
OTUD5	300713	301056	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked	80,636
OTUD6B	612021	617452	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	97,630
OTX2	600037	610125	Microphthalmia, syndromic 5	99,674
P4HTM	614584	618493	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities	92,382
PACS1	607492	615009	Schuurs-Hoeijmakers syndrome	93,392
PACS2	610423	618067	Developmental and epileptic encephalopathy 66	95,934
PAFAH1B1	601545	607432	Lissencephaly 1	99,164
PAH	612349	261600	Phenylketonuria	99,654
PAK3	300142	300558	Mental retardation, X-linked 30/47	91,208
PANK2	606157	234200	Neurodegeneration with brain iron accumulation 1	90,348
PAX1	167411	615560	Otofaciocervical syndrome 2	84,511
PAX3	606597	122880	Craniofacial-deafness-hand syndrome	99,683
PAX6	607108	106210	Aniridia	99,560
PC	608786	266150	Pyruvate carboxylase deficiency	99,686
PCDH12	605622	251280	Diencephalic-mesencephalic junction dysplasia syndrome 1	99,687
PCDH19	300460	300088	Epileptic encephalopathy, early infantile, 9	99,658
PCGF2	600346	618371	Turnpenny-Fry syndrome	99,688
PCNT	605925	210720	Microcephalic osteodysplastic primordial dwarfism, type II	99,463
PCYT2	602679	615033	Spastic paraplegia 82, autosomal recessive	92,078
PDE4D	600129	614613	Acrodysostosis 2, with or without hormone resistance	95,391
PDE6D	602676	615665	Joubert syndrome 22	99,643
PDHA1	300502	312170	Pyruvate dehydrogenase E1-alpha deficiency	97,669
PDHB	179060	614111	Pyruvate dehydrogenase E1-beta deficiency	99,477
PDHX	608769	245349	Lacticacidemia due to PDX1 deficiency	98,719
PDP1	605993	608782	Pyruvate dehydrogenase phosphatase deficiency	99,415
PDSS1	607429	614651	Coenzyme Q10 deficiency, primary, 2	85,774
PDSS2	610564	614652	Coenzyme Q10 deficiency, primary, 3	99,177
PEPD	613230	170100	Prolidase deficiency	99,652
PET100	614770	619055	Mitochondrial complex IV deficiency, nuclear type 12	99,688
PEX1	602136	214100	Peroxisome biogenesis disorder 1A (Zellweger)	98,989
PEX10	602859	614870	Peroxisome biogenesis disorder 6A (Zellweger)	88,693
PEX11B	603867	614920	Peroxisome biogenesis disorder 14B	99,624
PEX12	601758	614859	Peroxisome biogenesis disorder 3A (Zellweger)	99,565
PEX13	601789	614883	Peroxisome biogenesis disorder 11A (Zellweger)	99,425
PEX14	601791	614887	Peroxisome biogenesis disorder 13A (Zellweger)	99,662
PEX16	603360	614876	Peroxisome biogenesis disorder 8A (Zellweger)	99,688
PEX19	600279	614886	Peroxisome biogenesis disorder 12A (Zellweger)	99,572
PEX2	170993	614866	Peroxisome biogenesis disorder 5A (Zellweger)	99,686
PEX26	608666	614872	Peroxisome biogenesis disorder 7A (Zellweger)	96,308
PEX3	603164	614882	Peroxisome biogenesis disorder 10A (Zellweger)	98,863
PEX5	600414	214110	Peroxisome biogenesis disorder 2A (Zellweger)	99,218
PEX6	601498	614862	Peroxisome biogenesis disorder 4A (Zellweger)	95,057
PEX7	601757	614879	Peroxisome biogenesis disorder 9B	88,422
PGAP1	611655	615802	Mental retardation, autosomal recessive 42	97,602
PGAP2	615187	614207	Hyperphosphatasia with mental retardation syndrome 3	99,678
PGAP3	611801	615716	Hyperphosphatasia with mental retardation syndrome 4	99,225
PGK1	311800	300653	Phosphoglycerate kinase 1 deficiency	99,254
PGM1	171900	614921	Congenital disorder of glycosylation, type It	99,122
PHC1	602978	615414	Microcephaly 11, primary, autosomal recessive	99,662
PHF6	300414	301900	Borjeson-Forssman-Lehmann syndrome	85,174
PHF8	300560	300263	Mental retardation syndrome, X-linked, Siderius type	99,223
PHGDH	606879	601815	Phosphoglycerate dehydrogenase deficiency	99,675
PHIP	612870	617991	Chung-Jansen syndrome	97,853
PIBF1	607532	617767	Joubert syndrome 33	97,571
PIGA	311770	300868	Multiple congenital anomalies-hypotonia-seizures syndrome 2	95,370
PIGB	604122	618580	Developmental and epileptic encephalopathy 80	99,174
PIGC	601730	601730	Glycosylphosphatidylinositol biosynthesis defect 16	99,672
PIGG	616918	616917	Mental retardation, autosomal recessive 53	99,550
PIGL	605947	280000	CHIME syndrome	99,586
PIGN	606097	614080	Multiple congenital anomalies-hypotonia-seizures syndrome 1	97,815
PIGO	614730	614749	Hyperphosphatasia with mental retardation syndrome 2	99,675
PIGQ	605754	618548	Developmental and epileptic encephalopathy 77	99,640
PIGT	610272	615398	Multiple congenital anomalies-hypotonia-seizures syndrome 3	99,631
PIGV	610274	239300	Hyperphosphatasia with mental retardation syndrome 1	99,674
PIGW	610275	616025	Hyperphosphatasia with mental retardation syndrome 5	99,680
PIGY	610662	616809	Hyperphosphatasia with mental retardation syndrome 6	99,664
PIK3R2	603157	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	88,275
PLA2G6	603604	610217	Neurodegeneration with brain iron accumulation 2B	99,686
PLCB1	607120	613722	Epileptic encephalopathy, early infantile, 12	99,195
PLCB4	600810	614669	Auriculocondylar syndrome 2	99,134
PLK4	605031	616171	Microcephaly and chorioretinopathy, autosomal recessive, 2	98,868
PLOD1	153454	225400	Ehlers-Danlos syndrome, type VI	98,818
PLP1	300401	312080	Pelizaeus-Merzbacher disease	99,841
PLPBP	604436	617290	Epilepsy, early-onset, vitamin B6-dependent	97,338
PMM2	601785	212065	Congenital disorder of glycosylation, type Ia	98,283
PNKP	605610	613402	Microcephaly, seizures, and developmental delay	99,675
PNP	164050	613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency	99,663
PNPO	603287	610090	Pyridoxamine 5'-phosphate oxidase deficiency	99,554
PNPT1	610316	614932	Combined oxidative phosphorylation deficiency 13	98,646
POC1A	614783	614813	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	99,507
POGZ	614787	616364	White-Sutton syndrome	99,189
POLA1	312040	301030	Van Esch-O'Driscoll syndrome	92,132
POLG	174763	613662	Mitochondrial DNA depletion syndrome 4B (MNGIE type)	99,489
POLH	603968	278750	Xeroderma pigmentosum, variant type	99,567
POLR2A	180660	618603	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities	98,293
POLR3A	614258	607694	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	99,413
POLR3B	614366	614381	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	99,294

POMGNT1	606822	613151	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	99,667
POMGNT2	614828	614830	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8	99,688
POMK	615247	615249	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	99,667
POMT1	607423	613155	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	99,573
POMT2	607439	613156	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	97,182
PORCN	300651	305600	Focal dermal hypoplasia	99,886
POU3F3	602480	-	Snijders Blok et al., AJHG 2019 : developmental delays and/or intellectual disability and impairments in speech and language skills	46,787
PPM1D	605100	617450	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold	97,719
PPP1CB	600590	617506	Noonan syndrome-like disorder with loose anagen hair 2	99,373
PPP2R1A	605983	616362	Mental retardation, autosomal dominant 36	99,686
PPP2R5D	601646	616355	Mental retardation, autosomal dominant 35	99,667
PPT1	600722	256730	Ceroid lipofuscinosis, neuronal, 1	99,672
PQBP1	300463	309500	Renpenning syndrome	99,904
PRICKLE1	608500	612437	Epilepsy, progressive myoclonic 1B	99,679
PRMT7	610087	617157	Short stature, brachydactyly, intellectual developmental disability, and seizures	99,217
PRPS1	311850	301835	Arts syndrome	98,637
PRRT2	614386	605751	Seizures, benign familial infantile, 2	99,688
PRSS12	606709	249500	Mental retardation, autosomal recessive 1	97,452
PSAP	176801	611722	Krabbe disease, atypical	99,675
PSAT1	610936	616038	Neu-Laxova syndrome 2	99,568
PSMD12	604450	617516	Stankiewicz-Isidor syndrome	99,440
PTCH1	601309	610828	Holoprosencephaly 7	97,530
PTCHD1	300828	300830	Autism, susceptibility to, X-linked 4	99,571
PTDSS1	612792	151050	Lenz-Majewski hyperostotic dwarfism	99,588
PTEN	601728	158350	Cowden syndrome 1	98,904
PTPN11	176876	151100	LEOPARD syndrome 1	98,852
PTRF	603198	613327	Lipodystrophy, congenital generalized, type 4	99,242
PTRH2	608625	616263	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	99,688
PTS	612719	261640	Hyperphenylalaninemia, BH4-deficient, A	97,599
PUF60	604819	615583	Verheij syndrome	99,646
PURA	600473	616158	Mental retardation, autosomal dominant 31	84,892
PUS1	608109	600462	Myopathy, lactic acidosis, and sideroblastic anemia 1	92,963
PYCR1	179035	612940	Cutis laxa, autosomal recessive, type IIB	99,680
QARS1	603727	615760	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	99,681
QDPR	612676	261630	Hyperphenylalaninemia, BH4-deficient, C	96,667
QRICH1	617387	617982	Ververi-Brady syndrome	99,666
RAB11B	604198	617807	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter	99,680
RAB18	602207	614222	Warburg micro syndrome 3	98,526
RAB23	606144	201000	Carpenter syndrome	98,664
RAB27A	603868	607624	Griscelli syndrome, type 2	95,592
RAB39B	300774	300271	Mental retardation, X-linked 72	99,946
RAB3GAP1	602536	600118	Warburg micro syndrome 1	99,255
RAB3GAP2	609275	212720	Martsolf syndrome	99,209
RAC1	602048	617751	Mental retardation, autosomal dominant 48	93,182
RAD21	606462	614701	Cornelia de Lange syndrome 4	99,357
RAF1	164760	611553	Noonan syndrome 5	99,660
RAI1	607642	182290	Smith-Magenis syndrome	99,627
RARB	180220	615524	Microphthalmia, syndromic 12	99,540
RARS1	107820	616140	Leukodystrophy, hypomyelinating, 9	97,975
RARS2	611524	611523	Pontocerebellar hypoplasia, type 6	99,227
RASA2	601589	-	Aoki et al., JHG 2016 : RASopathie	94,842
RAX	601881	611038	Microphthalmia, isolated 3	75,564
RBBP8	604124	606744	Seckel syndrome 2	97,370
RBM10	300080	311900	TARP syndrome	99,880
RBM28	612074	612079	Alopecia, neurologic defects, and endocrinopathy syndrome	99,567
RBM8A	605313	274000	Thrombocytopenia-absent radius syndrome	99,679
RBPJ	147183	614814	Adams-Oliver syndrome 3	99,504
RECQL4	603780	218600	Baller-Gerold syndrome	95,511
RELN	600514	257320	Lissencephaly 2 (Norman-Roberts type)	99,510
RERE	605226	616975	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	99,556
RFT1	611908	612015	Congenital disorder of glycosylation, type In	96,745
RHOBTB2	607352	618004	Developmental and epileptic encephalopathy 64	99,669
RIT1	609591	615355	Noonan syndrome 8	99,660
RMND1	614917	614922	Combined oxidative phosphorylation deficiency 11	98,991
RNASEH2A	606034	610333	Aicardi-Goutieres syndrome 4	99,688
RNASEH2B	610326	610181	Aicardi-Goutieres syndrome 2	91,590
RNASEH2C	610330	610329	Aicardi-Goutieres syndrome 3	95,803
RNASET2	612944	612951	Leukoencephalopathy, cystic, without megalencephaly	99,472
RNF113A	300951	300953	Trichothiodystrophy 5, nonphotosensitive	99,854
ROGDI	614574	226750	Kohlschutter-Tonz syndrome	92,873
RORB	601972	618357	{Epilepsy, idiopathic generalized, susceptibility to, 15}	99,434
RPGRIP1L	610937	611560	Joubert syndrome 7	95,738
RPIA	180430	608611	Ribose 5-phosphate isomerase deficiency	96,036
RPL10	312173	300998	Mental retardation, X-linked, syndromic, 35	99,603
RPS6KA3	300075	303600	Coffin-Lowry syndrome	86,684
RRAS	165090	-	Flex et al., HMG 2014 : RASopathie	92,861
RRAS2	600098	618624	Noonan syndrome 12	88,699
RRM2B	604712	612075	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	98,685
RSRC1	613352	618402	Intellectual developmental disorder, autosomal recessive 70	96,224
RTEL1	608833	615190	Dyskeratosis congenita, autosomal recessive 5	99,685
RTN4IP1	610502	616732	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	99,530
RTTN	610436	614833	Microcephaly, short stature, and polymicrogyria with seizures	98,811
RXYLT1	605862	615041	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	96,390
SALL1	602218	107480	Townes-Brocks branchiootorenal-like syndrome	99,686
SAMHD1	606754	612952	Aicardi-Goutieres syndrome 5	99,499
SATB1	602075	619228	Developmental delay with dysmorphic facies and dental anomalies	99,565
SATB2	608148	612313	Glass syndrome	99,553
SC5D	602286	607330	Lathosterolosis	99,063
SCARB2	602257	254900	Epilepsy, progressive myoclonic 4, with or without renal failure	99,451
SCN1A	182389	607208	Epileptic encephalopathy, early infantile, 6 (Dravet syndrome)	99,084
SCN1B	600235	604233	Epilepsy, generalized, with febrile seizures plus, type 1	91,176
SCN2A	182390	613721	Epileptic encephalopathy, early infantile, 11	99,040
SCN3A	182391	617938	Developmental and epileptic encephalopathy 62	99,292
SCN8A	600702	614558	Epileptic encephalopathy, early infantile, 13	99,480
SCO2	604272	604377	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	99,683
SDCCAG8	613524	615993	Bardet-Biedl syndrome 16	99,215
SDHAF1	612848	252011	Mitochondrial complex II deficiency	66,110
SEPSECS	613009	613811	Pontocerebellar hypoplasia type 2D	99,109
SERAC1	614725	614739	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	99,004
SETBP1	611060	616078	Mental retardation, autosomal dominant 29	98,836
SETD2	612778	616831	Luscan-Lumish syndrome	99,474
SETD5	615743	615761	Mental retardation, autosomal dominant 23	99,581
SGSH	605270	252900	Mucopolysaccharidosis type IIIA (Sanfilippo A)	97,963
SHANK2	603290	613436	Autism susceptibility 17	96,795
SHANK3	606230	606232	Phelan-McDermid syndrome	73,964
SHH	600725	142945	Holoprosencephaly 3	83,252
SHOC2	602775	607721	Noonan-like syndrome with loose anagen hair	98,320
SHROOM4	300579	300434	Stocco dos Santos X-linked mental retardation syndrome	99,815
SIK1	605705	616341	Developmental and epileptic encephalopathy 30	99,624
SIL1	608005	248800	Marinesco-Sjogren syndrome	99,661
SIN3A	607776	613406	Witteveen-Kolk syndrome	99,575
SIX3	603714	157170	Holoprosencephaly 2	91,508
SKI	164780	182212	Shprintzen-Goldberg syndrome	87,894
SLC12A5	606726	616645	Developmental and epileptic encephalopathy 34	99,568
SLC12A6	604878	218000	Agenesis of the corpus callosum with peripheral neuropathy	99,206
SLC13A5	608305	615905	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta	99,666
SLC16A2	300095	300523	Allan-Herndon-Dudley syndrome	99,655
SLC17A5	604322	604369	Salla disease	98,558
SLC19A3	606152	607483	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	99,566
SLC1A2	600300	617105	Developmental and epileptic encephalopathy 41	99,462
SLC1A4	600229	616657	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	94,146
SLC25A1	190315	615182	Combined D-2- and L-2-hydroxyglutaric aciduria	86,909
SLC25A12	603667	612949	Epileptic encephalopathy, early infantile, 39	99,462
SLC25A15	603861	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	99,566
SLC25A19	606521	607196	Microcephaly, Amish type	99,666
SLC25A22	609302	609304	Epileptic encephalopathy, early infantile, 3	99,683
SLC2A1	138140	606777	GLUT1 deficiency syndrome 1, infantile onset, severe	98,015
SLC33A1	603690	614482	Congenital cataracts, hearing loss, and neurodegeneration	98,985
SLC35A1	605634	603585	Congenital disorder of glycosylation, type IIf	99,135

SLC35A2	314375	300896	Congenital disorder of glycosylation, type IIm	99,989
SLC35A3	605632	615553	?Arthrogryposis, mental retardation, and seizures	98,167
SLC35C1	605881	266265	Congenital disorder of glycosylation, type IIc	99,688
SLC39A8	608732	616721	Congenital disorder of glycosylation, type IIn	97,708
SLC45A1	605763	617532	Intellectual developmental disorder with neuropsychiatric features	97,089
SLC46A1	611672	229050	Folate malabsorption, hereditary	97,953
SLC4A4	603345	604278	Renal tubular acidosis, proximal, with ocular abnormalities	99,309
SLC6A1	137165	616421	Myoclonic-atonic epilepsy	99,609
SLC6A17	610299	616269	Mental retardation, autosomal recessive 48	99,681
SLC6A3	126455	613135	Parkinsonism-dystonia, infantile	99,681
SLC6A5	604159	614618	Hyperekplexia 3	99,109
SLC6A8	300036	300352	Cerebral creatine deficiency syndrome 1	88,971
SLC7A7	603593	222700	Lysinuric protein intolerance	99,676
SLC9A6	300231	300243	Mental retardation, X-linked syndromic, Christianson type	83,699
SMARCA2	600014	601358	Nicolaides-Baraitser syndrome	96,788
SMARCA4	603254	614609	Coffin-Siris syndrome 4	99,679
SMARCB1	601607	614608	Coffin-Siris syndrome 3	99,516
SMARCE1	603111	616938	Coffin-Siris syndrome 5	99,316
SMC1A	300040	300590	Cornelia de Lange syndrome 2	97,862
SMC3	606062	610759	Cornelia de Lange syndrome 3	98,625
SMOC1	608488	206920	Microphthalmia with limb anomalies	99,039
SMPD1	607608	257200	Niemann-Pick disease, type A	99,683
SMS	300105	309583	Mental retardation, X-linked, Snyder-Robinson type	87,751
SNAP25	600322	-	Klöckner et al., Genet Med 2021 : early-onset developmental and epileptic encephalopathy	99,567
SNAP29	604202	609528	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	98,861
SNIP1	608241	614501	Psychomotor retardation, epilepsy, and craniofacial dysmorphism	99,611
SNX14	616105	616354	Spinocerebellar ataxia, autosomal recessive 20	98,041
SOBP	613667	613671	Mental retardation, anterior maxillary protrusion, and strabismus	84,807
SON	182465	617140	ZTTK syndrome	99,454
SOS1	182530	610733	Noonan syndrome 4	98,391
SOS2	601247	616559	Noonan syndrome 9	98,649
SOX10	602229	611584	Waardenburg syndrome, type 2E, with or without neurologic involvement	93,767
SOX11	600898	615866	Mental retardation, autosomal dominant, 27	84,369
SOX2	184429	206900	Optic nerve hypoplasia and abnormalities of the central nervous system	95,936
SOX5	604975	616803	Lamb-Shaffer syndrome	99,582
SPATA5	613940	616577	Epilepsy, hearing loss, and mental retardation syndrome	99,194
SPECC1L	614140	145410	Opitz GBBB syndrome, type II	99,549
SPR	182125	612716	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	73,832
SPRED1	609291	611431	Legius syndrome	99,388
SPTAN1	182810	613477	Epileptic encephalopathy, early infantile, 5	99,489
SPTBN4	606214	617519	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	88,091
SRCAP	611421	136140	Floating-Harbor syndrome	99,664
SRD5A3	611715	612379	Congenital disorder of glycosylation, type Iq	96,475
ST3GAL3	606494	611090	Mental retardation, autosomal recessive 12	99,665
ST3GAL5	604402	609056	Salt and pepper developmental regression syndrome	92,000
STAG1	604358	617635	Mental retardation, autosomal dominant 47	98,331
STAG2	300826	301043	Holoprosencephaly 13, X-linked	88,125
STAMBP	606247	614261	Microcephaly-capillary malformation syndrome	99,614
STIL	181590	612703	Microcephaly 7, primary, autosomal recessive	99,369
STRA6	610745	601186	Microphthalmia, syndromic 9	99,674
STRADA	608626	611087	Polyhydramnios, megalencephaly, and symptomatic epilepsy	99,538
STT3A	601134	615596	Congenital disorder of glycosylation, type Iw	99,552
STT3B	608605	615597	Congenital disorder of glycosylation, type Ix	92,350
STX1B	601485	616172	Generalized epilepsy with febrile seizures plus, type 9	99,685
STXBP1	602926	612164	Epileptic encephalopathy, early infantile, 4	99,606
SUCLA2	603921	612073	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	99,264
SUCLG1	611224	245400	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	98,249
SUMF1	607939	272200	Multiple sulfatase deficiency	99,434
SUOX	606887	272300	Sulfite oxidase deficiency	99,686
SURF1	185620	256000	Leigh syndrome, due to COX IV deficiency	91,581
SUZ12	606245	618786	Imagawa-Matsumoto syndrome	95,903
SVBP	617853	-	Iqbal et al., Genet Med 2019	99,674
SYN1	313440	300491	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	74,291
SYNCRIP	616686	-	Lelieveld et al., Nat Neurosci 2016 : ID	99,477
SYNE1	608441	610743	Spinocerebellar ataxia, autosomal recessive 8	99,481
SYNGAP1	603384	612621	Mental retardation, autosomal dominant 5	99,684
SYNJ1	604297	617389	Developmental and epileptic encephalopathy 53	98,458
SYP	313475	300802	Mental retardation, X-linked 96	97,568
SYT14	610949	614229	Spinocerebellar ataxia, autosomal recessive 11	92,164
SZT2	615463	615476	Epileptic encephalopathy, early infantile, 18	99,667
TACO1	612958	619052	Mitochondrial complex IV deficiency, nuclear type 8	95,213
TAF1	313650	300966	Mental retardation, X-linked, syndromic 33	97,649
TAF2	604912	615599	Mental retardation, autosomal recessive 40	98,748
TAOK1	610266	-	Van Woerden et al., Hum Mutat 2021 : developmental delay/intellectual disability and/or variable learning or behavioral problems, muscular hypotonia, infant feeding difficulties, and growth problems.	99,249
TBC1D20	611663	615663	Warburg micro syndrome 4	92,920
TBC1D23	617687	617695	Pontocerebellar hypoplasia, type 11	97,486
TBC1D24	613577	615338	Epileptic encephalopathy, early infantile, 16	99,661
TBC1D7	612655	248000	Macrocephaly/megalencephaly syndrome, autosomal recessive	98,934
TBCD	604649	617193	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	96,683
TBCE	604934	241410	Hypoparathyroidism-retardation-dysmorphism syndrome	98,415
TBCK	616899	616900	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	98,328
TBL1XR1	608628	616944	Mental retardation, autosomal dominant 41	98,532
TBR1	604616	606053	Intellectual developmental disorder with autism and speech delay	87,671
TCF20	603107	618430	Developmental delay with variable intellectual impairment and behavioral abnormalities	99,680
TCF4	602272	610954	Pitt-Hopkins syndrome	99,654
TCOF1	606847	154500	Treacher Collins syndrome 1	99,621
TCTN1	609863	614173	Joubert syndrome 13	99,257
TCTN2	613846	616654	Joubert syndrome 24	99,089
TCTN3	613847	614815	Joubert syndrome 18	99,611
TECPR2	615000	615031	Spastic paraplegia 49, autosomal recessive	99,345
TECR	610057	614020	Mental retardation, autosomal recessive 14	99,688
TFAP2A	107580	113620	Branchiooculofacial syndrome	99,540
TGIF1	602630	142946	Holoprosencephaly 4	99,688
TH	191290	605407	Segawa syndrome, recessive	98,594
THOC2	300395	300957	Mental retardation, X-linked 12/35	91,526
THOC6	615403	613680	Beaulieu-Boycott-Innes syndrome	99,655
THRA	190120	614450	Hypothyroidism, congenital, nongoitrous, 6	99,581
TIMM8A	300356	304700	Mohr-Tranebjaerg syndrome	99,657
TINF2	604319	613990	Dyskeratosis congenita, autosomal dominant 3	99,661
TLK2	608439	618050	Mental retardation, autosomal dominant 57	99,174
TMCO1	614123	213980	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	95,627
TMEM107	616183	617563	Orofaciodigital syndrome XVI	99,688
TMEM138	614459	614465	Joubert syndrome 16	99,681
TMEM165	614726	614727	Congenital disorder of glycosylation, type IIk	84,395
TMEM216	613277	608091	Joubert syndrome 2	99,688
TMEM231	614949	614970	Joubert syndrome 20	99,625
TMEM237	614423	614424	Joubert syndrome 14	98,123
TMEM67	609884	610688	Joubert syndrome 6	97,767
TMEM70	612418	614052	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	97,387
TNK2	606994	-	Hitomi et al., Ann Neurol 2013 : severe autosomal recessive infantile-onset epilepsy and ID	99,401
TOGARAM1	617618	619185	Joubert syndrome 37	98,803
TPK1	606370	614458	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	99,014
TPP1	607998	609270	Spinocerebellar ataxia, autosomal recessive 7	99,640
TPP2	190470	619220	Immunodeficiency 78 with autoimmunity and developmental delay	98,476
TRAPPC11	614138	615356	Muscular dystrophy, limb-girdle, type 2S	98,780
TRAPPC4	610971	618741	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	99,688
TRAPPC9	611966	613192	Mental retardation, autosomal recessive 13	99,566
TREX1	606609	225750	Aicardi-Goutieres syndrome 1, dominant and recessive	99,688
TRIM32	602290	615988	Bardet-Biedl syndrome 11	99,685
TRIM8	606125	-	McClatchey et al., EJHG 2020 : Focal segmental glomerulosclerosis and mild intellectual disability	99,177
TRIO	601893	617061	Mental retardation, autosomal dominant 44	95,265
TRIP12	604506	617752	Mental retardation, autosomal dominant 49	99,281
TRMT10A	616013	616033	Microcephaly, short stature, and impaired glucose metabolism 1	99,099
TRMU	610230	613070	Liver failure, transient infantile	95,592
TRPM6	607009	602014	Hypomagnesemia 1, intestinal	99,450
TRPV4	605427	184252	Spondylometaphyseal dysplasia, Kozlowski type	99,650
TRRAP	603015	618454	Developmental delay with or without dysmorphic facies and autism	99,494
TSEN2	608753	612389	Pontocerebellar hypoplasia type 2B	99,625
TSEN54	608755	277470	Pontocerebellar hypoplasia type 2A	91,486

TSFM	604723	610505	Combined oxidative phosphorylation deficiency 3	92,099
TSPAN7	300096	300210	Mental retardation, X-linked 58	98,906
TTC19	613814	615157	Mitochondrial complex III deficiency, nuclear type 2	82,872
TTC37	614589	222470	Trichohepatoenteric syndrome 1	99,198
TTC8	608132	615985	Bardet-Biedl syndrome 8	98,883
TTI2	614426	615541	Mental retardation, autosomal recessive 39	99,599
TUBA1A	602529	611603	Lissencephaly 3	99,683
TUBA8	605742	613180	Cortical dysplasia, complex, with other brain malformations 8	99,424
TUBB	191130	615771	Cortical dysplasia, complex, with other brain malformations 6	99,688
TUBB2A	615101	615763	Cortical dysplasia, complex, with other brain malformations 5	98,614
TUBB2B	612850	610031	Cortical dysplasia, complex, with other brain malformations 7	99,685
TUBB3	602661	614039	Cortical dysplasia, complex, with other brain malformations 1	98,529
TUBB4A	602662	612438	Leukodystrophy, hypomyelinating, 6	99,372
TUBG1	191135	615412	Cortical dysplasia, complex, with other brain malformations 4	99,688
TUBGCP6	610053	251270	Microcephaly and chorioretinopathy, autosomal recessive, 1	99,686
TUSC3	601385	611093	Mental retardation, autosomal recessive 7	99,428
TWIST1	601622	101400	Saethre-Chotzen syndrome	59,507
TWNK	606075	609286	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3	99,619
UBA5	610552	617132	Developmental and epileptic encephalopathy 44	98,447
UBE2A	312180	300860	Mental retardation, X-linked syndromic, Nascimento-type	81,712
UBE3A	601623	105830	Angelman syndrome	99,004
UBE3B	608047	244450	Kaufman oculocerebrofacial syndrome	99,438
UBR1	605981	243800	Johanson-Blizzard syndrome	99,093
UFC1	610554	618076	Neurodevelopmental disorder with spasticity and poor growth	99,129
UFM1	610553	617899	Leukodystrophy, hypomyelinating, 14	98,714
UNC80	612636	616801	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	99,498
UPF3B	300298	300676	Mental retardation, X-linked, syndromic 14	92,065
UQCRQ	612080	615159	Mitochondrial complex III deficiency, nuclear type 4	99,632
UROC1	613012	276880	Urocanase deficiency	99,688
USP18	607057	617397	Pseudo-TORCH syndrome 2	99,685
USP9X	300072	300919	Mental retardation, X-linked 99	94,558
VARS2	612802	615917	Combined oxidative phosphorylation deficiency 20	99,661
VLDLR	192977	224050	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	97,915
VPS13B	607817	216550	Cohen syndrome	99,007
VPS4A	609982	619273	CIMDAG syndrome	98,351
VPS53	615850	615851	Pontocerebellar hypoplasia, type 2E	99,503
VRK1	602168	607596	Pontocerebellar hypoplasia type 1A	99,033
WAC	615049	616708	Desanto-Shinawi syndrome	98,130
WDPCP	613580	615992	Bardet-Biedl syndrome 15	98,981
WDR26	617424	617616	Skraban-Deardorff syndrome	99,491
WDR37	618586	618652	Neurooculocardiogenitourinary syndrome	99,531
WDR45	300526	300894	Neurodegeneration with brain iron accumulation 5	99,940
WDR45B	609226	617977	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	99,053
WDR62	613583	604317	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	99,590
WDR73	616144	251300	Galloway-Mowat syndrome	99,566
WDR81	614218	610185	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	99,488
WWOX	605131	616211	Epileptic encephalopathy, early infantile, 28	99,640
XPA	611153	278700	Xeroderma pigmentosum, group A	94,653
XPC	613208	278720	Xeroderma pigmentosum, group C	99,662
XYLT1	608124	615777	Desbuquois dysplasia 2	87,580
YAP1	606608	120433	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation	92,618
YWHAG	605356	617665	Developmental and epileptic encephalopathy 56	99,686
YY1	600013	617557	Gabriele-de Vries syndrome	94,574
ZBTB16	176797	612447	Skeletal defects, genital hypoplasia, and mental retardation	99,687
ZBTB18	608433	612337	Mental retardation, autosomal dominant 22	98,541
ZBTB20	606025	259050	Primrose syndrome	99,678
ZBTB24	614064	614069	Immunodeficiency-centromeric instability-facial anomalies syndrome-2	99,665
ZC4H2	300897	314580	Wieacker-Wolff syndrome	98,039
ZDHHC9	300646	300799	Mental retardation, X-linked syndromic, Raymond type	98,871
ZEB2	605802	235730	Mowat-Wilson syndrome	99,027
ZFYVE26	612012	270700	Spastic paraplegia 15, autosomal recessive	99,543
ZIC1	600470	618736	Structural brain anomalies with impaired intellectual development and craniosynostosis	99,183
ZIC2	603073	609637	Holoprosencephaly 5	61,706
ZMPSTE24	606480	608612	Mandibuloacral dysplasia with type B lipodystrophy	98,691
ZMYND11	608668	616083	Mental retardation, autosomal dominant 30	99,477
ZNF335	610827	615095	Microcephaly 10, primary, autosomal recessive	99,619
ZNF423	604557	614844	Joubert syndrome 19	99,680
ZNF711	314990	300803	Mental retardation, X-linked 97	95,682
ZSWIM6	615951	617865	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features	86,517

La nomenclature des gènes suit les recommandations HGCN : Genomics 79(4):464-470 (2002) [updated October 2013].
Les codes OMIM (gene, disease, phenotype) affichés ont été repris à partir de la version datant du 2019-10-23
La couverture moyenne avec une profondeur de lecture de 30x [Length Covered Above 30x] est calculée sur base de 320 exomes.

Gene symbols used follow HGCN guidelines : Genomics 79(4):464-470 (2002) [updated October 2013].
OMIM release used for OMIM disease identifiers and descriptions : 2019-10-23
[Length Covered Above 30x] describes the average coverage of a gene across 320 exomes in percentile.