CHU Liège : Modes de Transmission [Panel - DI v5 (1091 genes)]

Gene (HGNC)	Transcript sans version	OMIM (gene)	OMIM (disease)	OMIM (phenotype)	Mode de transmission	CI	AD	AR	XLD	XLR	Digenic	Onco	Mitochondrial	Mosaicism	Imprinting	Response to treatment
AAAS	NM_015665	605378	231550	Achalasia-addisonianism-alacrimia syndrome	AR			X								
AARS1	NM_001605	601065	616339	Developmental and epileptic encephalopathy 29	AR			X								
ABAT	NM_020686	137150	613163	GABA-transaminase deficiency	AR			X								
ABCC9	NM_005691	601439	239850	Hypertrichotic osteochondrodysplasia	AD		X									
ABCD1	NM_000033	300371	300100	Adrenoleukodystrophy	XLR					X						
ABHD5	NM_016006	604780	275630	Chanarin-Dorfman syndrome	AR			X								
ACAD9	NM_014049	611103	611126	Mitochondrial complex I deficiency due to ACAD9 deficiency	AR			X								
ACO2	NM_001098	100850	614559	Infantile cerebellar-retinal degeneration	AR			X								
ACOX1	NM_004035	609751	264470	Peroxisomal acyl-CoA oxidase deficiency	AR			X								
ACSF3	NM_174917	614245	614265	Combined malonic and methylmalonic aciduria	AR			X								
ACSL4	NM_004458	300157	300387	Mental retardation, X-linked 63	XLD				X							
ACTB	NM_001101	102630	243310	Baraitser-Winter syndrome 1	AD		X									
ACTG1	NM_001614	102560	614583	Baraitser-Winter syndrome 2	AD		X									
ACTL6B	NM_016188	612458	618468	Developmental and epileptic encephalopathy 76	AR			X								
ACY1	NM_000666	104620	609924	Aminoacylase 1 deficiency	AR			X								
ADAMTSL2	NM_014694	612277	231050	Geleophysic dysplasia 1	AR			X								
ADAR	NM_001111	146920	615010	Aicardi-Goutieres syndrome 6	AR		-	X								
ADAT3	NM_138422	615302	615286	Mental retardation, autosomal recessive 36	AR			X								
ADGRG1	NM_005682	604110	606854	Polymicrogyria, bilateral frontoparietal	AR			X								
ADK	NM_001123	102750	614300	Hypermethioninemia due to adenosine kinase deficiency	AR			X								
ADNP	NM_015339	611386	615873	Helsmoortel-van der Aa syndrome	AD		X									
ADSL	NM_000026	608222	103050	Adenylosuccinase deficiency	AR			X								
AFF2	NM_002025	300806	309548	Mental retardation, X-linked, FRAXE type	XLR					X						
AFF3	NM_002285	601464	619297	KINSSHIP syndrome	AD		X									
AFF4	NM_014423	604417	616368	CHOPS syndrome	AD		X									
AGA	NM_000027	613228	208400	Aspartylglucosaminuria	AR			X								
AGPAT2	NM_006412	603100	608594	Lipodystrophy, congenital generalized, type 1	AR			X								
AGPS	NM_003659	603051	600121	Rhizomelic chondrodysplasia punctata, type 3	AR			X								
AGTPBP1	NM_001286715	606830	618276	Neurodegeneration, childhood-onset, with cerebellar atrophy	AR			X								
AHCY	NM_000687	180960	613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AR			X								
AHDC1	NM_001029882	615790	615829	Xia-Gibbs syndrome	AD		X									
AHI1	NM_017651	608894	608629	Joubert syndrome 3	AR			X								
AIFM1	NM_004208	300169	300816	Combined oxidative phosphorylation deficiency 6	XLR					X						
AIMP1	NM_004757	603605	260600	Leukodystrophy, hypomyelinating, 3	AR			X								
AKT3	NM_005465	611223	615937	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	AD		X							X		
ALDH18A1	NM_002860	138250	219150	Cutis laxa, autosomal recessive, type IIIA	CI	X	X	X								
ALDH3A2	NM_000382	609523	270200	Sjogren-Larsson syndrome	AR			X								
ALDH4A1	NM_003748	606811	239510	Hyperprolinemia, type II	AR			X								
ALDH5A1	NM_001080	610045	271980	Succinic semialdehyde dehydrogenase deficiency	AR			X								
ALDH7A1	NM_001182	107323	266100	Epilepsy, pyridoxine-dependent	AR			X								
ALG1	NM_019109	605907	608540	Congenital disorder of glycosylation, type Ik	AR			X								
ALG11	NM_001004127	613666	613661	Congenital disorder of glycosylation, type Ip	AR			X								
ALG12	NM_024105	607144	607143	Congenital disorder of glycosylation, type Ig	AR			X								
ALG13	NM_001099922	300776	300884	Epileptic encephalopathy, early infantile, 36	XLD				X							
ALG2	NM_033087	607905	616228	Myasthenic syndrome, congenital, 14, with tubular aggregates	AR			X								
ALG3	NM_005787	608750	601110	Congenital disorder of glycosylation, type Id	AR			X								
ALG6	NM_013339	604566	603147	Congenital disorder of glycosylation, type Ic	AR			X								
ALG8	NM_024079	608103	608104	Congenital disorder of glycosylation, type Ih	AR			X								
ALG9	NM_024740	606941	263210	Gillessen-Kaesbach-Nishimura syndrome	AR			X								
ALMS1	NM_015120	606844	203800	Alstrom syndrome	AR			X								
ALX1	NM_006982	601527	613456	Frontonasal dysplasia 3	AR			X								
ALX4	NM_021926	605420	613451	Frontonasal dysplasia 2	CI	X	X	X								
AMER1	NM_152424	300647	300373	Osteopathia striata with cranial sclerosis	XLD				X							
AMPD2	NM_001368809	102771	615809	Pontocerebellar hypoplasia, type 9	AR			X								
AMT	NM_000481	238310	605899	Glycine encephalopathy	AR			X								
ANK3	NM_020987	600465	615493	Mental retardation, autosomal recessive, 37	CI	X	X	X								
ANKH	NM_054027	605145	123000	Craniometaphyseal dysplasia	AD		X									
ANKRD11	NM_013275	611192	148050	KBG syndrome	AD		X									
AP1S1	NM_001283	603531	609313	MEDNIK syndrome	AR			X								
AP1S2	NM_003916	300629	304340	Mental retardation, X-linked syndromic 5	XLR					X						
AP3B2	NM_004644	602166	617276	Developmental and epileptic encephalopathy 48	AR			X								
AP4B1	NM_006594	607245	614066	Spastic paraplegia 47, autosomal recessive	AR			X								
AP4E1	NM_007347	607244	614744	Spastic paraplegia 51, autosomal recessive	AR			X								
AP4M1	NM_004722	602296	612936	Spastic paraplegia 50, autosomal recessive	AR			X								
AP4S1	NM_007077	607243	614067	Spastic paraplegia 52, autosomal recessive	AR			X								
APTX	NM_175073	606350	208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	AR			X								
ARCN1	NM_001655	600820	617164	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay	AD		X									
ARFGEF2	NM_006420	605371	608097	Periventricular heterotopia with microcephaly	AR			X								
ARG1	NM_000045	608313	207800	Argininemia	AR			X								
ARHGAP31	NM_020754	610911	100300	Adams-Oliver syndrome 1	AD		X									
ARHGEF2	NM_004723	607560	617523	Neurodevelopmental disorder with midbrain and hindbrain malformations	AR			X								
ARHGEF9	NM_015185	300429	300607	Epileptic encephalopathy, early infantile, 8	XLR					X						

ARID1A	NM_006015	603024	614607	Coffin-Siris syndrome 2	AD		X									
ARID1B	NM_020732	614556	135900	Coffin-Siris syndrome 1	AD		X									
ARID2	NM_152641	609539	617808	Coffis-Siris syndrome 6	AD		X									
ARL13B	NM_182896	608922	612291	Joubert syndrome 8	AR			X								
ARL6	NM_177976	608845	600151	Bardet-Biedl syndrome 3	AR			X			X					
ARSA	NM_000487	607574	250100	Metachromatic leukodystrophy	AR			X								
ARSL	NM_000047	300180	302950	Chondrodysplasia punctata, X-linked recessive	XLR					X						
ARX	NM_139058	300382	300419	Mental retardation, X-linked 29 and others	XLR					X						
ASAH1	NM_177924	613468	159950	Spinal muscular atrophy with progressive myoclonic epilepsy	AR			X								
ASH1L	NM_018489	607999	617796	Mental retardation, autosomal dominant 52	AD		X									
ASL	NM_000048	608310	207900	Argininosuccinic aciduria	AR			X								
ASNS	NM_133436	108370	615574	Asparagine synthetase deficiency	AR			X								
ASPA	NM_000049	608034	271900	Canavan disease	AR			X								
ASPM	NM_018136	605481	608716	Microcephaly 5, primary, autosomal recessive	AR			X								
ASS1	NM_000050	603470	215700	Citrullinemia	AR			X								
ASXL1	NM_015338	612990	605039	Bohring-Opitz syndrome	AD		X									
ASXL3	NM_030632	615115	615485	Bainbridge-Ropers syndrome	AD		X									
ATIC	NM_004044	601731	608688	AICA-ribosiduria due to ATIC deficiency	AR			X								
ATN1	NM_001007026	607462	618494	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies	AD		X									
ATP1A2	NM_000702	182340	602481	Migraine, familial hemiplegic, 2	AD		X									
ATP1A3	NM_152296	182350	601338	CAPOS syndrome	AD		X									
ATP5F1A	NM_001001937	164360	615228	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4	AR			X								
ATP6AP2	NM_005765	300556	300423	Mental retardation, X-linked, syndromic, Hedera type	XLR					X						
ATP6V0A2	NM_012463	611716	219200	Cutis laxa, autosomal recessive, type IIA	AR			X								
ATP6V1A	NM_001690	607027	618012	Developmental and epileptic encephalopathy 93	AD		X									
ATP7A	NM_000052	300011	309400	Menkes disease	XLR					X						
ATPAF2	NM_145691	608918	604273	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	AR			X								
ATR	NM_001184	601215	210600	Seckel syndrome 1	AR		-	X				X				
ATRIP	NM_130384	606605	-	Ogi et al., PLoS Genet 2012 : Seckel syndrome	AR			X								
ATRX	NM_000489	300032	309580	Mental retardation-hypotonic facies syndrome, X-linked	XLR				-	X						
AUH	NM_001698	600529	250950	3-methylglutaconic aciduria, type I	AR			X								
AUTS2	NM_015570	607270	615834	Mental retardation, autosomal dominant 26	AD		X									
B3GALNT2	NM_152490	610194	615181	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	AR			X								
B3GLCT	NM_194318	610308	261540	Peters-plus syndrome	AR			X								
B4GALT1	NM_001497	137060	607091	Congenital disorder of glycosylation, type IId	AR			X								
B4GALT7	NM_007255	604327	130070	Ehlers-Danlos syndrome with short stature and limb anomalies	AR			X								
B4GAT1	NM_006876	605517	615287	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	AR			X								
BBIP1	NM_001195306	613605	615995	Bardet-Biedl syndrome 18	AR			X								
BBS1	NM_024649	209901	209900	Bardet-Biedl syndrome 1	AR			X			X					
BBS10	NM_024685	610148	615987	Bardet-Biedl syndrome 10	AR			X								
BBS12	NM_152618	610683	615989	Bardet-Biedl syndrome 12	AR			X								
BBS2	NM_031885	606151	615981	Bardet-Biedl syndrome 2	AR			X								
BBS4	NM_033028	600374	615982	Bardet-Biedl syndrome 4	AR			X								
BBS5	NM_152384	603650	615983	Bardet-Biedl syndrome 5	AR			X								
BBS7	NM_176824	607590	615984	Bardet-Biedl syndrome 7	AR			X								
BBS9	NM_198428	615986	615986	Bardet-Biedl syndrome 9	AR			X								
BCAP31	NM_001139441	300398	300475	Deafness, dystonia, and cerebral hypomyelination	XLR					X						
BCKDHA	NM_000709	608348	248600	Maple syrup urine disease, type Ia	AR			X								
BCKDHB	NM_183050	248611	248600	Maple syrup urine disease, type Ib	AR			X								
BCKDK	NM_005881	614901	614923	Branched-chain ketoacid dehydrogenase kinase deficiency	AR			X								
BCL11A	NM_022893	606557	617101	Dias-Logan syndrome	AD		X									
BCOR	NM_017745	300485	300166	Microphthalmia, syndromic 2	XLD				X							
BCS1L	NM_004328	603647	256000	Leigh syndrome	AR			X					X			
BLM	NM_000057	210900	210900	Bloom syndrome	AR			X				X				
BOLA3	NM_212552	613183	614299	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	AR			X								
BRAF	NM_004333	164757	613706	Noonan syndrome 7	AD		X					X				
BRAT1	NM_152743	614506	618056	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	AR			X								
BRF1	NM_001519	604902	616202	Cerebellofaciodental syndrome	AR			X								
BRPF1	NM_001003694	602410	617333	Intellectual developmental disorder with dysmorphic facies and ptosis	AD		X									
BRSK2	NM_001256627	609236	-	Hiatt et al., AJHG 2019	AD		X									
BRWD3	NM_153252	300553	300659	Mental retardation, X-linked 93	XLR					X						
BSCL2	NM_032667	606158	615924	Encephalopathy, progressive, with or without lipodystrophy	AR		-	X								
BTD	NM_001370658	609019	253260	Biotinidase deficiency	AR			X								
BUB1B	NM_001211	602860	257300	Mosaic variegated aneuploidy syndrome 1	AR			X								
c12orf4	NM_020374	616082	618221	Mental retardation, autosomal recessive 66	AR			X								Philips et al., Clin Genet 2017 : mild
C12ORF57	NM_138425	615640	218340	Temtamy syndrome	AR			X								
C12orf65	NM_152269	613541	615035	Spastic paraplegia 55, autosomal recessive	AR			X								
C19ORF12	NM_001031726	614297	614298	Neurodegeneration with brain iron accumulation 4	AR			X								
C2CD3	NM_015531	615944	615948	Orofaciodigital syndrome XIV	AR			X								
C5ORF42	NM_023073	614571	614615	Joubert syndrome 17	AR			X								
CA8	NM_004056	114815	613227	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	AR			X								
CACNA1A	NM_001127221	601011	617106	Epileptic encephalopathy, early infantile, 42	AD		X									
CACNA1C	NM_000719	114205	601005	Timothy syndrome	AD		X									
CACNA1D	NM_000720	114206	615474	Primary aldosteronism, seizures, and neurologic abnormalities	AD		X	X								
CACNA1E	NM_000721	601013	618285	Developmental and epileptic encephalopathy 69	AD		X									

CACNA2D2	NM_006030	607082	618501	Cerebellar atrophy with seizures and variable developmental delay	AR			X								
CAD	NM_004341	114010	616457	Developmental and epileptic encephalopathy 50	AR			X								
CAMK2A	NM_015981	114078	617798	Mental retardation, autosomal dominant 53	AD		X									
CAMK2B	NM_001220	607707	617799	Mental retardation, autosomal dominant 54	AD		X									
CANT1	NM_138793	613165	251450	Desbuquois dysplasia 1	AR			X								
CASK	NM_003688	300172	300749	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	XLD				X							
CAV1	NM_001753	601047	606721	Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome	AD		X									
CBL	NM_005188	165360	613563	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	AD		X						X			
CBS	NM_000071	613381	236200	Homocystinuria, B6-responsive and nonresponsive types	AR			X								
CC2D1A	NM_017721	610055	608443	Mental retardation, autosomal recessive 3	AR			X								
CC2D2A	NM_001080522	612013	612285	Joubert syndrome 9	AR			X								
CCBE1	NM_133459	612753	235510	Hennekam lymphangiectasia-lymphedema syndrome 1	AR			X								
CCDC22	NM_014008	300859	300963	Ritscher-Schinzel syndrome 2	XLR					X						
CCDC39	NM_181426	613798	613807	Ciliary dyskinesia, primary, 14	AR			X								
CCDC78	NM_001031737	614666	614807	Myopathy, centronuclear, 4	AD		X									
CCDC88C	NM_001080414	611204	236600	Hydrocephalus, nonsyndromic, autosomal recessive	AR		-	X								
CCND2	NM_001759	123833	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3	AD		X									
CD96	NM_198196	606037	211750	C syndrome	AR			X								
CDC42	NM_001791	116952	616737	Takenouchi-Kosaki syndrome	AD		X									
CDC45	NM_001178010	603465	617063	Meier-Gorlin syndrome 7	AR			X								
CDC6	NM_001254	602627	613805	Meier-Gorlin syndrome 5	AR			X								
CDH15	NM_004933	114019	612580	Mental retardation, autosomal dominant 3	AD		X									
CDK10	NM_052988	603464	617694	Al Kaissi syndrome	AR			X								
CDK13	NM_003718	603309	617360	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	AD		X									
CDK5RAP2	NM_018249	608201	604804	Microcephaly 3, primary, autosomal recessive	AR			X								
CDK8	NM_001260	603184	612100	Intellectual developmental disorder with hypotonia and behavioral abnormalities	AD		X									
CDKL5	NM_003159	300203	300672	Epileptic encephalopathy, early infantile, 2	XLD				X						X	
CDON	NM_016952	608707	614226	Holoprosencephaly 11	AD		X									
CDT1	NM_030928	605525	613804	Meier-Gorlin syndrome 4	AR			X								
CENPJ	NM_018451	609279	608393	Microcephaly 6, primary, autosomal recessive	AR			X								
CEP104	NM_014704	616690	614615	Joubert syndrome 25	AR			X								
CEP135	NM_025009	611423	614673	Microcephaly 8, primary, autosomal recessive	AR			X								
CEP152	NM_014985	613529	614852	Microcephaly 9, primary, autosomal recessive	AR			X								
CEP290	NM_025114	610142	610188	Joubert syndrome 5	AR			X								
CEP41	NM_018718	610523	614464	Joubert syndrome 15	AR			X								
CEP57	NM_014679	607951	614114	Mosaic variegated aneuploidy syndrome 2	AR			X								
CEP63	NM_025180	614724	614728	Seckel syndrome 6	AR			X								
CHAMP1	NM_001164144	616327	616579	Mental retardation, autosomal dominant 40	AD		X									
CHAT	NM_020549	118490	254210	Myasthenic syndrome, congenital, 6, presynaptic	AR			X								
CHD2	NM_001271	602119	615369	Epileptic encephalopathy, childhood-onset	AD		X									
CHD4	NM_001273	603277	617159	Sifrim-Hitz-Weiss syndrome	AD		X									
CHD7	NM_017780	608892	214800	CHARGE syndrome	AD		X									
CHD8	NM_001170629	610528	615032	Autism, susceptibility to, 18	AD		X									
CHKB	NM_005198	612395	602541	Muscular dystrophy, congenital, megaconial type	AR			X								
CHMP1A	NM_002768	164010	614961	Pontocerebellar hypoplasia, type 8	AR			X								
CHRNA2	NM_000742	118502	610253	Epilepsy, nocturnal frontal lobe, type 4	AD		X									
CHRNA4	NM_000744	118504	600513	Epilepsy, nocturnal frontal lobe, 1	AD		X									
CHRNB2	NM_000748	118507	605375	Epilepsy, nocturnal frontal lobe, 3	AD		X									
CHRND	NM_000751	100720	616322	Myasthenic syndrome, congenital, 3B, fast-channel	AR		-	X								
CISD2	NM_001008388	611507	604928	Wolfram syndrome 2	AR			X								
CLCN2	NM_004366	600570	607628	{Epilepsy, juvenile myoclonic, susceptibility to, 8}	CI	X	X	X								
CLCN4	NM_001830	302910	300114	Mental retardation, X-linked 49/15	XLR					X						
CLN3	NM_001042432	607042	204200	Ceroid lipofuscinosis, neuronal, 3	AR			X								
CLN5	NM_006493	608102	256731	Ceroid lipofuscinosis, neuronal, 5	AR			X								
CLN6	NM_017882	606725	601780	Ceroid lipofuscinosis, neuronal, 6	AR			X								
CLN8	NM_018941	607837	600143	Ceroid lipofuscinosis, neuronal, 8	AR			X								
CLP1	NM_006831	608757	615803	Pontocerebellar hypoplasia, type 10	AR			X								
CLPB	NM_030813	616254	616271	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	AR			X								
CLTC	NM_004859	118955	617854	Mental retardation, autosomal dominant 56	AD		X									
CNKSR2	NM_014927	300724	301008	Mental retardation, X-linked, syndromic, Houge type	XLD				X							
CNNM2	NM_017649	607803	616418	Hypomagnesemia, seizures, and mental retardation	AR			X								
CNOT3	NM_014516	604910	618672	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies	AD		X									
CNTNAP1	NM_003632	602346	616286	Lethal congenital contracture syndrome 7	AR			X								
CNTNAP2	NM_014141	604569	610042	Pitt-Hopkins like syndrome 1	AR			X								
COA8	NM_001370595	616003	220110	Mitochondrial complex IV deficiency, nuclear type 17	AR			X								
COASY	NM_025233	609855	615643	Neurodegeneration with brain iron accumulation 6	AR			X								
COG1	NM_018714	606973	611209	Congenital disorder of glycosylation, type IIg	AR			X								
COG4	NM_015386	606976	613489	Congenital disorder of glycosylation, type IIj	CI	X	X	X								
COG5	NM_006348	606821	613612	Congenital disorder of glycosylation, type IIi	AR			X								
COG6	NM_020751	606977	614576	Congenital disorder of glycosylation, type III	AR			X								
COG7	NM_153603	606978	608779	Congenital disorder of glycosylation, type IIe	AR			X								
COG8	NM_032382	606979	611182	Congenital disorder of glycosylation, type IIh	AR			X								
COL4A1	NM_001845	120130	175780	Porencephaly 1	AD		X									
COL4A2	NM_001846	120090	614483	Porencephaly 2	AD		X									
COL4A3BP	NM_001130105	604677	616351	Mental retardation, autosomal dominant 34	AD		X									

COLEC10	NM_006438	607620	616351	Mental retardation, autosomal dominant 34	AR			X								
COLEC11	NM_024027	612502	265050	3MC syndrome 2	AR			X								
COQ2	NM_015697	609825	607426	Coenzyme Q10 deficiency, primary, 1	AR			X								
COQ4	NM_016035	612898	616276	Coenzyme Q10 deficiency, primary, 7	AR			X								
COQ6	NM_182476	614647	614650	Coenzyme Q10 deficiency, primary, 6	AR			X								
COQ7	NM_016138	601683	616733	Coenzyme Q10 deficiency, primary, 8	AR			X								
COQ8A	NM_020247	606980	612016	Coenzyme Q10 deficiency, primary, 4	AR			X								
COQ9	NM_020312	612837	614654	Coenzyme Q10 deficiency, primary, 5	AR			X								
COX10	NM_001303	602125	256000	Leigh syndrome due to mitochondrial COX4 deficiency	AR			X					X			
COX15	NM_004376	603646	615119	Mitochondrial complex IV deficiency, nuclear type 6	AR			X								
CPS1	NM_001875	608307	237300	Carbamoylphosphate synthetase I deficiency	AR			X								
CRADD	NM_003805	603454	614499	Mental retardation, autosomal recessive 34, with variant lissencephaly	AR			X								
CRBN	NM_016302	609262	607417	Mental retardation, autosomal recessive 2	AR			X								
CREBBP	NM_004380	600140	180849	Rubinstein-Taybi syndrome 1	AD		X									
CRPPA	NM_001101426	614631	614643	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	AR			X								
CSNK2A1	NM_001895	115440	617062	Okur-Chung neurodevelopmental syndrome	AD		X									
CSPP1	NM_024790	611654	615636	Joubert syndrome 21	AR			X								
CSTB	NM_000100	601145	254800	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	AR			X								
CTC1	NM_025099	613129	612199	Cerebroretinal microangiopathy with calcifications and cysts	AR			X								
CTCF	NM_006565	604167	615502	Mental retardation, autosomal dominant 21	AD		X									
CTNNB1	NM_001904	116806	615075	Mental retardation, autosomal dominant 19	AD		X									
CTSA	NM_000308	613111	256540	Galactosialidosis	AR			X								
CTSD	NM_001909	116840	610127	Ceroid lipofuscinosis, neuronal, 10	AR			X								
CUL3	NM_003590	603136	619239	Neurodevelopmental disorder with or without autism or seizures	AD		X									
CUL4B	NM_003588	300304	300354	Mental retardation, X-linked, syndromic 15 (Cabezas type)	XLR					X						
CWF19L1	NM_018294	616120	616127	Spinocerebellar ataxia, autosomal recessive 17	AR			X								
CYB5R3	NM_000398	613213	250800	Methemoglobinemia, type I	AR			X								
CYFIP2	NM_001037333	606323	618468	Developmental and epileptic encephalopathy 65	AD		X									
CYP27A1	NM_000784	606530	213700	Cerebrotendinous xanthomatosis	AR			X								
CYP2U1	NM_183075	610670	615030	Spastic paraplegia 56, autosomal recessive	AR			X								
D2HGDH	NM_152783	609186	600721	D-2-hydroxyglutaric aciduria	AR			X								
DAG1	NM_004393	128239	616538	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9	AR			X								
DARS1	NM_001349	603084	615281	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	AR			X								
DARS2	NM_018122	610956	611105	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	AR			X								
DBT	NM_001918	248610	248600	Maple syrup urine disease, type II	AR			X								
DCAF17	NM_025000	612515	241080	Woodhouse-Sakati syndrome	AR			X								
DCC	NM_005215	120470	157600	Mirror movements 1 and/or agenesis of the corpus callosum	CI	X	X	X				X				
DCPS	NM_014026	610534	616459	Al-Raqad syndrome	AR			X								
DCX	NM_178153	300121	300067	Lissencephaly, X-linked	XLD				X							
DDB2	NM_000107	600811	278740	Xeroderma pigmentosum, group E, DDB-negative subtype	AR			X								
DDC	NM_000790	107930	608643	Aromatic L-amino acid decarboxylase deficiency	AR			X								
DDHD2	NM_015214	615003	615033	Spastic paraplegia 54, autosomal recessive	AR			X								
DDX11	NM_030653	601150	613398	Warsaw breakage syndrome	AR			X								
DDX3X	NM_001193416	300160	300958	Mental retardation, X-linked 102	XLD				X							
DEAF1	NM_021008	602635	615828	Mental retardation, autosomal dominant 24	CI	X	X	X								
DEGS1	NM_003676	615843	618404	Leukodystrophy, hypomyelinating, 18	AR			X								
DEPDC5	NM_001242896	614191	604364	Epilepsy, familial focal, with variable foci 1	AD		X									
DHCR24	NM_014762	606418	602398	Desmosterolosis	AR			X								
DHCR7	NM_001360	602858	270400	Smith-Lemli-Opitz syndrome	AR			X								
DHDDS	NM_024887	608172	617836	Developmental delay and seizures with or without movement abnormalities	AD		X									
DHFR	NM_000791	126060	613839	Megaloblastic anemia due to dihydrofolate reductase deficiency	AR			X								
DHX30	NM_138615	616423	617804	Neurodevelopmental disorder with severe motor impairment and absent language	AD		X									
DIAPH1	NM_005219	602121	616632	Seizures, cortical blindness, microcephaly syndrome	CI	X	X	X								
DKC1	NM_001363	300126	305000	Dyskeratosis congenita, X-linked	XLR					X						
DLD	NM_000108	238331	246900	Dihydrolipoamide dehydrogenase deficiency	AR			X								
DLG3	NM_021120	300189	300850	Mental retardation, X-linked 90	XLR					X						
DLL4	NM_019074	605185	616589	Adams-Oliver syndrome 6	AD		X									
DNAJC19	NM_145261	608977	610198	3-methylglutaconic aciduria, type V	AR			X								
DNM1	NM_004408	602377	616346	Epileptic encephalopathy, early infantile, 31	AD		X									
DNMT3A	NM_175629	602769	615879	Tatton-Brown-Rahman syndrome	AD		X									
DNMT3B	NM_006892	602900	242860	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	AR			X								
DOCK6	NM_020812	614194	614219	Adams-Oliver syndrome 2	AR			X								
DOCK7	NM_001271999	615730	615859	Developmental and epileptic encephalopathy 23	AR			X								
DOCK8	NM_203447	611432	243700	Hyper-IgE recurrent infection syndrome, autosomal recessive	AR			X								
DOLK	NM_014908	610746	610768	Congenital disorder of glycosylation, type Im	AR			X								
DPAGT1	NM_001382	191350	608093	Congenital disorder of glycosylation, type Ij	AR			X								
DPH1	NM_001383	603527	616901	Developmental delay with short stature, dysmorphic features, and sparse hair	AR			X								
DPM1	NM_003859	603503	608799	Congenital disorder of glycosylation, type Ie	AR			X								
DPM2	NM_003863	603564	615042	Congenital disorder of glycosylation, type Iu	AR			X								
DPP6	NM_001936	126141	616311	Mental retardation, autosomal dominant 33	AD		X									
DSCAM	NM_001389	602523	-	Wang et al., Nat Commun 2016 : Autisme	AD		X									
DYNC1H1	NM_001376	600112	614563	Mental retardation, autosomal dominant 13	AD		X									
DYRK1A	NM_001396	600855	614104	Mental retardation, autosomal dominant 7	AD		X									
EARS2	NM_001083614	612799	614924	Combined oxidative phosphorylation deficiency 12	AR			X								
EBF3	NM_001005463	607407	617330	Hypotonia, ataxia, and delayed development syndrome	AD		X									

EBP	NM_006579	300205	300960	MEND syndrome	CI	X			X	X						
ECHS1	NM_004092	602292	616277	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	AR			X								
EDC3	NM_025083	609842	616460	Mental retardation, autosomal recessive 50	AR			X								
EEF1A2	NM_001958	602959	616393	Mental retardation, autosomal dominant 38	AD		X									
EFNB1	NM_004429	300035	304110	Craniofrontonasal dysplasia	XLD				X							
EFTUD2	NM_004247	603892	610536	Mandibulofacial dysostosis, Guion-Almeida type	AD		X									
EHMT1	NM_024757	607001	610253	Kleefstra syndrome	AD		X									
EIF2AK3	NM_004836	604032	226980	Wolcott-Rallison syndrome	AR			X								
EIF2B1	NM_001414	606686	603896	Leukoencephalopathy with vanishing white matter	AR			X								
EIF2B2	NM_014239	606454	603897	Leukoencephalopathy with vanishing white matter	AR			X								
EIF2B3	NM_020365	606273	603898	Leukoencephalopathy with vanishing white matter	AR			X								
EIF2B4	NM_015636	606687	603899	Leukoencephalopathy with vanishing white matter	AR			X								
EIF2B5	NM_003907	603945	603900	Leukoencephalopathy with vanishing white matter	AR			X								
EIF2S3	NM_001415	300161	300987	Mental retardation, X-linked, syndromic, Borck type	XLR					X						
EIF3F	NM_003754	603914	618295	Mental retardation, autosomal recessive 67	AR			X								
ELAC2	NM_018127	605367	615440	Combined oxidative phosphorylation deficiency 17	AR			X								
ELOVL4	NM_022726	605512	614457	Ichthyosis, spastic quadriplegia, and mental retardation	CI	X	X	X								
ELP1	NM_003640	603722	223900	Dysautonomia, familial	AR			X								
ELP2	NM_001242875	616054	617270	Mental retardation, autosomal recessive 58	AR			X								
EML1	NM_004434	602033	600348	Band heterotopia	AR			X								
EMX2	NM_004098	600035	269160	Schizencephaly	AD		X									
ENTPD1	NM_001776	601752	615683	Spastic paraplegia 64, autosomal recessive	AR			X								
EOGT	NM_001278689	614789	615297	Adams-Oliver syndrome 4	AR			X								
EP300	NM_001429	602700	613684	Rubinstein-Taybi syndrome 2	AD		X									
EPB41L1	NM_012156	602879	614257	Mental retardation, autosomal dominant 11	AD		X									
EPG5	NM_020964	615068	242840	Vici syndrome	AR			X								
EPM2A	NM_005670	607566	254780	Epilepsy, progressive myoclonic 2A (Lafora)	AR			X								
ERCC2	NM_000400	126340	610756	Cerebrooculofacioskeletal syndrome 2	AR			X								
ERCC3	NM_000122	133510	610651	Xeroderma pigmentosum, group B	AR			X								
ERCC4	NM_005236	133520	278760	Xeroderma pigmentosum, type F/Cockayne syndrome	AR			X								
ERCC5	NM_000123	133530	278780	Xeroderma pigmentosum, group G/Cockayne syndrome	AR			X								
ERCC6	NM_000124	609413	214150	Cerebrooculofacioskeletal syndrome 1	AR			X								
ERCC8	NM_000082	609412	216400	Cockayne syndrome, type A	AR			X								
ERLIN2	NM_007175	611605	611225	Spastic paraplegia 18, autosomal recessive	AR			X								
ESCO2	NM_001017420	609353	268300	Roberts syndrome	AR			X								
ETHE1	NM_014297	608451	602473	Ethylmalonic encephalopathy	AR			X								
EXOSC3	NM_016042	606489	614678	Pontocerebellar hypoplasia, type 1B	AR			X								
EZH2	NM_004456	601573	277590	Weaver syndrome	AD		X									
EZR	NM_003379	123900	-	Riecken et al., Hum Mut 2015 : ID	AR			X								
FA2H	NM_024306	611026	612319	Spastic paraplegia 35, autosomal recessive	AR			X								
FAM126A	NM_032581	610531	610532	Leukodystrophy, hypomyelinating, 5	AR			X								
FARS2	NM_006567	611592	614946	Combined oxidative phosphorylation deficiency 14	AR			X								
FASTKD2	NM_014929	612322	618855	Combined oxidative phosphorylation deficiency 44	AR			X								
FAT4	NM_024582	612411	615546	Van Maldergem syndrome 2	AR			X								
FBXL4	NM_012160	605654	615471	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	AR			X								
FBXO11	NM_001190274	607871	618089	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities	AD		X									
FBXO31	NM_024735	609102	615979	Mental retardation, autosomal recessive 45	AR			X								
FGD1	NM_004463	300546	305400	Mental retardation, X-linked syndromic 16	XLR					X						
FGF12	NM_021032	601513	617166	Developmental and epileptic encephalopathy 47	AD		X									
FGF14	NM_004115	601515	609307	Spinocerebellar ataxia 27	AD		X									
FGFR1	NM_023110	136350	147950	Hypogonadotropic hypogonadism 2 with or without anosmia	AD		X									
FGFR2	NM_000141	176943	101400	Saethre-Chotzen syndrome	AD		X	-					X			
FGFR3	NM_000142	134934	612247	Crouzon syndrome with acanthosis nigricans	AD		X	-					X		X	
FH	NM_000143	136850	606812	Fumarase deficiency	CI	X	X	X					X			
FKRP	NM_024301	606596	606612	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5	AR			X								
FKTN	NM_001079802	607440	613152	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4	AR			X								
FLNA	NM_001456	300017	300049	Heterotopia, periventricular	CI	X			X	X						
FMN2	NM_020066	606373	616193	Mental retardation, autosomal recessive 47	AR			X								
FMR1	NM_002024	309550	300623	Fragile X tremor/ataxia syndrome	XLD				X							
FOLR1	NM_016725	136430	613068	Neurodegeneration due to cerebral folate transport deficiency	AR			X								
FOXG1	NM_005249	164874	613454	Rett syndrome, congenital variant	AD		X									
FOXP1	NM_032682	605515	613670	Mental retardation with language impairment and with or without autistic features	AD		X									
FOXP2	NM_014491	605317	602081	Speech-language disorder-1	AD		X									
FOXRED1	NM_017547	613622	256000	Leigh syndrome due to mitochondrial complex I deficiency	AR			X						X		
FRAS1	NM_025074	607830	219000	Fraser syndrome 1	AR			X								
FREM2	NM_207361	608945	617666	Fraser syndrome 2	AR			X								
FRMPD4	NM_014728	300838	300983	Mental retardation, X-linked 104	XLR					X						
FTCD	NM_006657	606806	229100	Glutamate formiminotransferase deficiency	AR			X								
FTL	NM_000146	134790	606159	Neurodegeneration with brain iron accumulation 3	AD		X	-								
FTO	NM_001080432	610966	612938	Growth retardation, developmental delay, facial dysmorphism	AR			X								
FTSJ1	NM_012280	300499	309549	Mental retardation, X-linked 9/44	XLR					X						
FUCA1	NM_000147	612280	230000	Fucosidosis	AR			X								
GABBR2	NM_005458	607340	617904	Developmental and epileptic encephalopathy 59	AD		X									
GABRA1	NM_000806	137160	615744	Developmental and epileptic encephalopathy 19	AD		X									
GABRA2	NM_001114175	137140	618557	Developmental and epileptic encephalopathy 78	AD		X									

GABRA3	NM_000808	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.	XLD					X							
GABRA5	NM_000810	137142	618559	Developmental and epileptic encephalopathy 79	AD			X									
GABRB2	NM_021911	600232	617829	Developmental and epileptic encephalopathy 92	AD			X									
GABRB3	NM_000814	137192	617113	Epileptic encephalopathy, early infantile, 43	AD			X									X
GABRG2	NM_000816	137164	611277	Epilepsy, generalized, with febrile seizures plus, type 3	AD			X								X	
GAD1	NM_000817	605363	619124	Developmental and epileptic encephalopathy 89	AR				X								
GALC	NM_000153	606890	245200	Krabbe disease	AR				X								
GALT	NM_000155	606999	230400	Galactosemia	AR				X								
GAMT	NM_000156	601240	612736	Cerebral creatine deficiency syndrome 2	AR				X								
GAN	NM_022041	605379	256850	Giant axonal neuropathy-1	AR				X								
GATAD2B	NM_020699	614998	615074	Mental retardation, autosomal dominant 18	AD			X									
GATM	NM_001482	602360	612718	Cerebral creatine deficiency syndrome 3	AR				X								
GBA2	NM_020944	609471	614409	Spastic paraplegia 46, autosomal recessive	AR				X								
GBE1	NM_000158	607839	232500	Glycogen storage disease IV	AR				X								
GCDH	NM_000159	608801	231670	Glutaricaciduria, type I	AR				X								
GCH1	NM_000161	600225	128230	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	CI	X	X		X								
GCSH	NM_004483	238330	605899	Glycine encephalopathy	AR				X								
GDI1	NM_001493	300104	300849	Mental retardation, X-linked 41	XLD					X							
GFAP	NM_002055	137780	203450	Alexander disease	AD			X									
GFM1	NM_024996	606639	609060	Combined oxidative phosphorylation deficiency 1	AR				X								
GJC2	NM_020435	608803	608804	Leukodystrophy, hypomyelinating, 2	CI	X	X		X								
GLB1	NM_000404	611458	230500	GM1-gangliosidosis, type I	AR				X								
GLDC	NM_000170	238300	605899	Glycine encephalopathy	AR				X								
GLI2	NM_005270	165230	610829	Holoprosencephaly 9	AD			X									
GLI3	NM_000168	165240	175700	Greig cephalopolysyndactyly syndrome	AD			X									
GLRA1	NM_000171	138491	149600	Hyperekplexia 1	CI	X	X		X								
GLRB	NM_000824	138492	614619	Hyperekplexia 2	AR				X								
GLUL	NM_002065	138290	610015	Glutamine deficiency, congenital	AR				X								
GLYCTK	NM_145262	610516	220120	D-glyceric aciduria	AR				X								
GM2A	NM_000405	613109	272750	GM2-gangliosidosis, AB variant	AR				X								
GMPPA	NM_205847	615495	615510	Alacrima, achalasia, and mental retardation syndrome	AR				X								
GMPPB	NM_013334	615320	615351	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14	AR				X								
GNAI3	NM_006496	139370	602483	Auriculocondylar syndrome 1	AD			X									
GNAO1	NM_020988	139311	615473	Epileptic encephalopathy, early infantile, 17	AD			X									
GNAS	NM_000516	139320	103580	Pseudohypoparathyroidism Ia	AD			X								X	
GNB1	NM_002074	139380	616973	Mental retardation, autosomal dominant 42	AD			X									
GNPAT	NM_014236	602744	222765	Rhizomelic chondrodysplasia punctata, type 2	AR				X								
GNPTAB	NM_024312	607840	252500	Mucolipidosis II alpha/beta	AR				X								
GNPTG	NM_032520	607838	252605	Mucolipidosis III gamma	AR				X								
GNS	NM_002076	607664	252940	Mucopolysaccharidosis type IIID	AR				X								
GOSR2	NM_004287	604027	614018	Epilepsy, progressive myoclonic 6	AR				X								
GPC3	NM_004484	300037	312870	Simpson-Golabi-Behmel syndrome, type 1	XLR						X						
GPHN	NM_020806	603930	615501	Molybdenum cofactor deficiency C	AR				X								
GPSM2	NM_013296	609245	604213	Chudley-McCullough syndrome	AR				X								
GPT2	NM_133443	138210	616281	Mental retardation, autosomal recessive 49	AR				X								
GRIA3	NM_000828	305915	300699	Mental retardation, X-linked 94	XLR						X						
GRIA4	NM_000829	138246	617864	Neurodevelopmental disorder with or without seizures and gait abnormalities	AD			X									
GRID2	NM_001510	602368	616204	Spinocerebellar ataxia, autosomal recessive 18	AR				X								
GRIK2	NM_021956	138244	611092	Mental retardation, autosomal recessive, 6	AR				X								
GRIN1	NM_007327	138249	614254	Mental retardation, autosomal dominant 8	AD			X	-								
GRIN2A	NM_000833	138253	245570	Epilepsy, focal, with speech disorder and with or without mental retardation	AD			X									
GRIN2B	NM_000834	138252	613970	Mental retardation, autosomal dominant 6	AD			X								X	
GRIN2D	NM_000836	602717	617162	Developmental and epileptic encephalopathy 46	AD			X									
GRIP1	NM_021150	604597	617667	Fraser syndrome 3	AR				X								
GRM1	NM_001278064	604473	617691	Spinocerebellar ataxia 44	AR			-	X								
GSS	NM_000178	601002	266130	Glutathione synthetase deficiency	AR				X								
GTF2E2	NM_002095	189964	616943	Trichothiodystrophy 6, nonphotosensitive	AR				X								
GTF2H5	NM_207118	608780	616395	Trichothiodystrophy 3, photosensitive	AR				X								
GTPBP2	NM_019096	607434	617988	Jaberi-Elahi syndrome	AR				X								
GTPBP3	NM_133644	608536	616198	Combined oxidative phosphorylation deficiency 23	AR				X								
GUSB	NM_000181	611499	253220	Mucopolysaccharidosis VII	AR				X								
HACE1	NM_020771	610876	616756	Spastic paraplegia and psychomotor retardation with or without seizures	AR				X								
HAX1	NM_006118	605998	610738	Neutropenia, severe congenital 3, autosomal recessive	AR				X					X			
HCFC1	NM_005334	300019	616756	Spastic paraplegia and psychomotor retardation with or without seizures	XLR						X						
HCN1	NM_021072	602780	615871	Epileptic encephalopathy, early infantile, 24	AD			X									
HDAC4	NM_006037	605314	-	Fisch et al., AJMG 2016 : ID	AD			X									
HDAC8	NM_018486	300269	300882	Cornelia de Lange syndrome 5	XLD					X							
HECW2	NM_020760	617245	617268	Neurodevelopmental disorder with hypotonia, seizures, and absent language	AD			X									
HEPACAM	NM_152722	611642	613926	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation	CI	X	X		X								
HERC1	NM_003922	605109	617011	Macrocephaly, dysmorphic facies, and psychomotor retardation	AR				X								
HERC2	NM_004667	605837	615516	Mental retardation, autosomal recessive 38	AR				X								
HESX1	NM_003865	601802	182230	Growth hormone deficiency with pituitary anomalies	CI	X	X		X								
HEXA	NM_000520	606869	272800	Tay-Sachs disease	AR				X								
HEXB	NM_000521	606873	268800	Sandhoff disease, infantile, juvenile, and adult forms	AR				X								
HGSNAT	NM_152419	610453	252930	Mucopolysaccharidosis type IIIC (Sanfilippo C)	AR				X								

HIBCH	NM_014362	610690	250620	3-hydroxyisobutryl-CoA hydrolase deficiency	AR			X								
HIVEP2	NM_006734	143054	616977	Mental retardation, autosomal dominant 43	AD		X							X		
HLCS	NM_000411	609018	253270	Holocarboxylase synthetase deficiency	AR			X								
HNMT	NM_006895	605238	616739	Mental retardation, autosomal recessive 51	AR			X								
HNRNPH2	NM_019597	300610	300986	Mental retardation, X-linked, syndromic, Bain type	XLD				X							
HNRNPK	NM_002140	600712	616580	Au-Kline syndrome	AD		X									
HNRNPU	NM_031844	602869	617391	Epileptic encephalopathy, early infantile, 54	AD		X									
HOXA1	NM_005522	142955	601536	Athabaskan brainstem dysgenesis syndrome	AR			X								
HPD	NM_002150	609695	276710	Tyrosinemia, type III	AR			X								
HPRT1	NM_000194	308000	300322	Lesch-Nyhan syndrome	XLR					X						
HRAS	NM_005343	190020	218040	Costello syndrome	AD		X						X		X	
HSD17B10	NM_004493	300256	300438	HSD10 mitochondrial disease	XLD				X							
HSPD1	NM_002156	118190	612233	Leukodystrophy, hypomyelinating, 4	AR			X								
HSPG2	NM_005529	142461	224410	Dyssegmental dysplasia, Silverman-Handmaker type	AR			X								
HUWE1	NM_031407	300697	300706	Mental retardation, X-linked syndromic, Turner type	XLD				X							
IARS1	NM_002161	600709	617093	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	AR			X								
IARS2	NM_018060	612801	616007	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	AR			X								
IDS	NM_000202	300823	309900	Mucopolysaccharidosis II	XLR					X						
IDUA	NM_000203	252800	607015	Mucopolysaccharidosis Ih/s	AR			X								
IER3IP1	NM_016097	609382	614231	Microcephaly, epilepsy, and diabetes syndrome	AR			X								
IFIH1	NM_022168	606951	615846	Aicardi-Goutieres syndrome 7	AD		X									
IFT172	NM_015662	607386	615630	Short-rib thoracic dysplasia 10 with or without polydactyly	AR			X								
IFT27	NM_006860	615870	615996	Bardet-Biedl syndrome 19	AR			X								
IFT57	NM_018010	606621	617927	?Orofaciodigital syndrome XVIII	AR			X								
IGF1	NM_000618	147440	608747	Growth retardation with deafness and mental retardation due to IGF1 deficiency	AR			X								
IL1RAPL1	NM_014271	300206	300143	Mental retardation, X-linked 21/34	XLR					X						
INPP5E	NM_019892	613037	213300	Joubert syndrome 1	AR			X								
INTU	NM_015693	610621	617926	?Orofaciodigital syndrome XVII	AR			X								
IQSEC2	NM_001111125	300522	309530	Mental retardation, X-linked 1/78	XLD				X							
IRX5	NM_005853	606195	611174	Hamamy syndrome	AR			X								
ITGA7	NM_002206	600536	613204	Muscular dystrophy, congenital, due to ITGA7 deficiency	AR			X								
ITPR1	NM_002222	147265	117360	Spinocerebellar ataxia 29, congenital nonprogressive	AD		X									
JAM3	NM_032801	606871	613730	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	AR			X								
KANSL1	NM_001193466	612452	610443	Koolen-De Vries syndrome	AD		X									
KAT6A	NM_006766	601408	616268	Mental retardation, autosomal dominant 32	AD		X									
KAT6B	NM_012330	605880	606170	Genitopatellar syndrome	AD		X									
KCNA1	NM_000217	176260	160120	Episodic ataxia/myokymia syndrome	AD		X									
KCNA2	NM_004974	176262	616366	Developmental and epileptic encephalopathy 32	AD		X									
KCNB1	NM_004975	600397	616056	Developmental and epileptic encephalopathy 26	AD		X									
KCNC1	NM_001112741	176258	616187	Epilepsy, progressive myoclonic 7	AD		X									
KCNC3	NM_004977	176264	605259	Spinocerebellar ataxia 13	AD		X									
KCNH1	NM_172362	603305	611816	Temple-Baraitser syndrome	AD		X									
KCNJ10	NM_002241	602208	612780	SESAME syndrome	AR			X								
KCNJ11	NM_000525	600937	606176	Diabetes, permanent neonatal, with or without neurologic features	AD		X	-								
KCNK9	NM_001282534	605874	612292	Birk-Barel mental retardation dysmorphism syndrome	AD		X								X	
KCNMA1	NM_002247	600150	609446	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy	AD		X									
KCNQ2	NM_172107	602235	613720	Epileptic encephalopathy, early infantile, 7	AD		X							X		
KCNQ3	NM_004519	602232	121201	Seizures, benign neonatal, 2	AD		X									
KCNQ5	NM_001160133	607357	617601	Mental retardation, autosomal dominant 46	AD		X									
KCNT1	NM_020822	608167	614959	Epileptic encephalopathy, early infantile, 14	AD		X									
KCNT2	NM_198503	610044	617771	Developmental and epileptic encephalopathy 57	AD		X									
KCTD7	NM_153033	611725	611726	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	AR			X								
KDM1A	NM_001009999	609132	616728	Cleft palate, psychomotor retardation, and distinctive facial features	AD		X									
KDM5C	NM_004187	314690	300534	Mental retardation, X-linked, syndromic, Claes-Jensen type	XLR					X						
KDM6A	NM_021140	300128	300867	Kabuki syndrome 2	XLD				X							
KIAA0586	NM_001244189	610178	616490	Joubert syndrome 23	AR			X								
KIAA0753	NM_014804	617112	617127	Orofaciodigital syndrome XV	AR			X								
KIAA2022	NM_001008537	300524	300912	Mental retardation, X-linked 98	XLD				X							
KIF11	NM_004523	148760	152950	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation	AD		X									
KIF14	NM_014875	611279	616258	Meckel syndrome 12	AR			X								
KIF1A	NM_004321	601255	614255	Mental retardation, autosomal dominant 9	CI	X	X	X								
KIF2A	NM_001098511	602591	615411	Cortical dysplasia, complex, with other brain malformations 3	AD		X									
KIF4A	NM_012310	300521	300923	Mental retardation, X-linked 100	XLR					X						
KIF5A	NM_004984	602821	617235	Myoclonus, intractable, neonatal	AD		X									
KIF5C	NM_004522	604593	615282	Cortical dysplasia, complex, with other brain malformations 2	AD		X									
KIF7	NM_198525	611254	200990	Joubert syndrome 12	AR			X								
KIFBP	NM_015634	609367	609460	Goldberg-Shprintzen megacolon syndrome	AR			X								
KIRREL3	NM_032531	607761	612581	Mental retardation, autosomal dominant 4	AD		X									
KLC2	NM_022822	611729	609541	Spastic paraplegia, optic atrophy, and neuropathy	AR			X								
KMT2A	NM_001197104	159555	605130	Wiedemann-Steiner syndrome	AD		X						X			
KMT2C	NM_170606	606833	617768	Kleefstra syndrome 2	AD		X									
KMT2D	NM_003482	602113	147920	Kabuki syndrome 1	AD		X							X		
KMT2E	NM_182931	608444	618512	O'Donnell-Luria-Rodan syndrome	AD		X									
KMT5B	NM_017635	610881	617788	Mental retardation, autosomal dominant 51	AD		X									
KNL1	NM_170589	609173	604321	Microcephaly 4, primary, autosomal recessive	AR			X								

KPTN	NM_007059	615620	615637	Mental retardation, autosomal recessive 41	AR			X								
KRAS	NM_004985	190070	609942	Noonan syndrome 3	AD		X					X		X		
L1CAM	NM_000425	308840	303350	CRASH syndrome	XLR					X						
L2HGDH	NM_024884	609584	236792	L-2-hydroxyglutaric aciduria	AR			X								
LAMA1	NM_005559	150320	615960	Poretti-Boltshauser syndrome	AR			X								
LAMA2	NM_000426	156225	607855	Muscular dystrophy, congenital, due to partial LAMA2 deficiency	AR			X								
LAMB1	NM_002291	150240	615191	Lissencephaly 5	AR			X								
LAMC3	NM_006059	604349	614115	Cortical malformations, occipital	AR			X								
LAMP2	NM_002294	309060	300257	Danon disease	XLD				X							
LARGE1	NM_004737	603590	608840	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	AR			X								
LARP7	NM_016648	612026	615071	Alazami syndrome	AR			X								
LAS1L	NM_031206	300964	309585	Wilson-Turner syndrome	XLR					X						
LIAS	NM_006859	607031	614462	Hyperglycinemia, lactic acidosis, and seizures	AR			X								
LIG4	NM_002312	601837	606593	LIG4 syndrome	AR			X								
LINS1	NM_001040616	610350	614340	Mental retardation, autosomal recessive 27	AR			X								
LIPT1	NM_145199	610284	616299	Lipoyltransferase 1 deficiency	AR			X								
LMBRD1	NM_018368	612625	277380	Methylmalonic aciduria and homocystinuria, cblF type	AR			X								
LMNA	NM_170707	150330	616516	Emery-Dreifuss muscular dystrophy 3, AR	CI	X	X	X								
LRP2	NM_004525	600073	222448	Donnai-Barrow syndrome	AR			X								
LRPPRC	NM_133259	607544	220111	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	AR			X								
LZTFL1	NM_020347	606568	615994	Bardet-Biedl syndrome 17	AR			X								
LZTR1	NM_006767	600574	616564	Noonan syndrome 10	AD		X									
MAF	NM_005360	177075	601088	Ayme-Gripp syndrome	AD		X									
MAGEL2	NM_019066	605283	615547	Schaaf-Yang syndrome	AD		X							X	X	
MAN1B1	NM_016219	604346	614202	Mental retardation, autosomal recessive 15	AR			X								
MAN2B1	NM_000528	609458	248500	Mannosidosis, alpha-, types I and II	AR			X								
MANBA	NM_005908	609489	248510	Mannosidosis, beta	AR			X								
MAOA	NM_000240	309850	300615	Brunner syndrome	XLR					X						
MAP2K1	NM_002755	176872	615279	Cardiofaciocutaneous syndrome 3	AD		X									
MAP2K2	NM_030662	601263	615280	Cardiofaciocutaneous syndrome 4	AD		X									
MASP1	NM_139125	600521	257920	3MC syndrome 1	AR			X								
MAT1A	NM_000429	610550	250850	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	CI	X	X	X								
MBD5	NM_018328	611472	156200	Mental retardation, autosomal dominant 1	AD		X									
MBOAT7	NM_024298	606048	617188	Mental retardation, autosomal recessive 57	AR			X								
MBTPS2	NM_015884	300294	308205	IFAP syndrome with or without BRESHECK syndrome	XLR					X						
MCCC1	NM_020166	609010	210200	3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR			X								
MCOLN1	NM_020533	605248	252650	Mucolipidosis IV	AR			X								
MCPH1	NM_024596	607117	251200	Microcephaly 1, primary, autosomal recessive	AR			X								
MECP2	NM_004992	300005	312750	Rett syndrome	XLD				X					X		
MED12	NM_005120	300188	305450	Opitz-Kaveggia syndrome	XLR					X						
MED13	NM_005121	603808	618009	Intellectual developmental disorder 61	AD		X									
~~MED13L~~	~~NM_015335~~	~~608771~~	~~616789~~	~~Mental retardation and distinctive facial features with or without cardiac defects~~	~~AD~~		~~X~~									
MED17	NM_004268	603810	613668	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR			X								
MED23	NM_015979	605042	614249	Mental retardation, autosomal recessive 18	AR			X								
MEF2C	NM_002397	600662	613443	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	AD		X									
MEIS2	NM_170674	601740	600987	Cleft palate, cardiac defects, and mental retardation	AD		X									
METTL23	NM_001080510	615262	615942	Mental retardation, autosomal recessive 44	AR			X								
MFF	NM_020194	614785	617086	Encephalopathy due to defective mitochondrial and peroxisomal fission 2	AR			X								
MFSD2A	NM_001136493	614397	616486	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	AR			X								
MFSD8	NM_152778	611124	610951	Ceroid lipofuscinosis, neuronal, 7	AR			X								
MGAT2	NM_002408	602616	212066	Congenital disorder of glycosylation, type IIa	AR			X								
MICU1	NM_006077	605084	615673	Myopathy with extrapyramidal signs	AR			X								
MID1	NM_000381	300552	300000	Opitz GBBB syndrome, type I	XLR					X						
MKKS	NM_018848	604896	605231	Bardet-Biedl syndrome 6	AR			X								
MKS1	NM_017777	609883	615990	Bardet-Biedl syndrome 13	AR			X								
MLC1	NM_015166	605908	604004	Megalencephalic leukoencephalopathy with subcortical cysts	AR			X								
MLYCD	NM_012213	606761	248360	Malonyl-CoA decarboxylase deficiency	AR			X								
MMACHC	NM_015506	609831	277400	Methylmalonic aciduria and homocystinuria, cblC type	AR			X								
MMADHC	NM_015702	611935	277410	Homocystinuria and Methylmalonic Aciduria	AR			X								
MOCS1	NM_005943	603707	252150	Molybdenum cofactor deficiency A	AR			X								
MOCS2	NM_176806	603708	252160	Molybdenum cofactor deficiency B	AR			X								
MOGS	NM_006302	601336	606056	Congenital disorder of glycosylation, type IIb	AR			X								
MORC2	NM_001303256	616661	619090	Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy	AD		X									
MPDU1	NM_004870	604041	609180	Congenital disorder of glycosylation, type If	AR			X								
MPDZ	NM_003829	603785	615219	Hydrocephalus, nonsyndromic, autosomal recessive 2	AR			X								
MPLKIP	NM_138701	609188	234050	Trichothiodystrophy 4, nonphotosensitive	AR			X								
MRAS	NM_012219	608435	618499	Noonan syndrome 11	AD		X									
MSL3	NM_001282174	300609	301032	Basilicata-Akhtar syndrome	XLD				X							
MTFMT	NM_139242	611766	614947	Combined oxidative phosphorylation deficiency 15	AR			X								
MTHFR	NM_005957	607093	236250	Homocystinuria due to MTHFR deficiency	AR			X								
MTM1	NM_000252	300415	310400	Myotubular myopathy, X-linked	XLR					X						
MTOR	NM_004958	601231	616638	Smith-Kingsmore syndrome	AD		X							X		
MTR	NM_000254	156570	250940	Homocystinuria-megaloblastic anemia, cblG complementation type	AR			X								
MTRR	NM_002454	602568	236270	Homocystinuria-megaloblastic anemia, cbl E type	AR			X								
MVK	NM_000431	251170	610377	Mevalonic aciduria	CI	X	X	X								

MYCN	NM_005378	164840	164280	Feingold syndrome 1	AD		X									
MYO5A	NM_000259	160777	214450	Griscelli syndrome, type 1	AR			X								
MYT1L	NM_015025	613084	616521	Mental retardation, autosomal dominant 39	AD		X									
NAA10	NM_003491	300013	300855	Ogden syndrome	CI	X			X	X						
NAA15	NM_057175	608000	617787	Mental retardation, autosomal dominant 50	AD		X									
NACC1	NM_052876	610672	617393	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination	AD		X									
NAGA	NM_000262	104170	609241	Schindler disease, type I	AR			X								
NAGLU	NM_000263	609701	252920	Mucopolysaccharidosis type IIIB (Sanfilippo B)	CI	X	X	X								
NALCN	NM_052867	611549	616266	Congenital contractures of the limbs and face, hypotonia, and developmental delay	CI	X	X	X								
NANS	NM_018946	605202	610442	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	AR			X								
NAPB	NM_001283018	611270	-	Conroy et al., Clin Genet 2016 : early-onset epileptic encephalopathy	AR			X								
NARS1	NM_004539	108410	619091	Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive	AR			X								
NARS2	NM_024678	612803	616239	Combined oxidative phosphorylation deficiency 24	AR			X								
NBN	NM_002485	602667	251260	Nijmegen breakage syndrome	AR			X								
NCDN	NM_001014839	608458	-	Fatima et al., AJHG 2021	CI	X	X	X								
NDE1	NM_001143979	609449	614019	Lissencephaly 4 (with microcephaly)	AR			X								
NDP	NM_000266	300658	310600	Norrie disease	XLR					X						
NDST1	NM_001543	600853	616116	Mental retardation, autosomal recessive 46	AR			X								
NDUFA1	NM_004541	300078	252010	Mitochondrial complex I deficiency	XLD				X				X			
NDUFA11	NM_175614	612638	252010	Mitochondrial complex I deficiency	AR			X					X			
NDUFA12	NM_018838	614530	256000	Leigh syndrome due to mitochondrial complex 1 deficiency	AR			X					X			
NDUFA2	NM_002488	602137	256000	Leigh syndrome due to mitochondrial complex I deficiency	AR			X					X			
NDUFA9	NM_005002	603834	256000	Leigh syndrome due to mitochondrial complex I deficiency	AR			X					X			
NDUFAF2	NM_174889	609653	618233	Mitochondrial complex I deficiency, nuclear type 10	AR			X								
NDUFAF3	NM_199069	612911	618240	Mitochondrial complex I deficiency, nuclear type 18	AR			X								
NDUFAF4	NM_014165	611776	618237	Mitochondrial complex I deficiency, nuclear type 15	AR			X								
NDUFAF5	NM_024120	612360	618238	Mitochondrial complex I deficiency, nuclear type 16	AR			X								
NDUFAF6	NM_152416	612392	256000	Leigh syndrome due to mitochondrial complex I deficiency	AR			X					X			
NDUFB3	NM_002491	603839	618246	Mitochondrial complex I deficiency, nuclear type 25	AR			X								
NDUFS1	NM_005006	157655	618226	Mitochondrial complex I deficiency, nuclear type 5	AR			X								
NDUFS2	NM_004550	602985	252010	Mitochondrial complex I deficiency	AR			X					X			
NDUFS3	NM_004551	603846	256000	Leigh syndrome due to mitochondrial complex I deficiency	AR			X					X			
NDUFS4	NM_002495	602694	252010	Mitochondrial complex I deficiency	AR			X					X			
NDUFS6	NM_004553	603848	618232	Mitochondrial complex I deficiency, nuclear type 9	AR			X								
NDUFS7	NM_024407	601825	256000	Leigh syndrome	AR			X					X			
NDUFS8	NM_002496	602141	618222	Mitochondrial complex I deficiency, nuclear type 2	AR			X								
NDUFV1	NM_007103	161015	618225	Mitochondrial complex I deficiency, nuclear type 4	AR			X								
NDUFV2	NM_021074	600532	618229	Mitochondrial complex I deficiency, nuclear type 7	AR			X								
NECAP1	NM_015509	611623	615833	Developmental and epileptic encephalopathy 21	AR			X								
NEDD4L	NM_015277	606384	617201	Periventricular nodular heterotopia 7	AD		X									
NEU1	NM_000434	608272	256550	Sialidosis	AR			X								
NFIA	NM_005595	600727	613735	Brain malformations and urinary tract defects	AD		X									
NFIX	NM_001271043	164005	614753	Sotos syndrome 2	AD		X									
NGF	NM_002506	162030	608654	Neuropathy, hereditary sensory and autonomic, type V	AR			X								
NGLY1	NM_018297	610661	615273	Congenital disorder of deglycosylation	AR			X								
NHEJ1	NM_024782	611290	611291	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	AR			X								
NHLRC1	NM_198586	608072	254780	Epilepsy, progressive myoclonic 2B (Lafora)	AR			X								
NHP2	NM_017838	606470	613987	Dyskeratosis congenita, autosomal recessive 2	AR			X								
NHS	NM_198270	300457	302350	Nance-Horan syndrome	XLD				X							
NIN	NM_020921	608684	614851	Seckel syndrome 7	AR			X								
NIPBL	NM_133433	608667	122470	Cornelia de Lange syndrome 1	AD		X									
NKAP	NM_024528	300766	301039	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type	XLR					X						
NKX2-1	NM_001079668	600635	610978	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	AD		X					X				
NONO	NM_001145408	300084	300967	Mental retardation, X-linked, syndromic 34	XLR					X						
NOTCH1	NM_017617	190198	616028	Adams-Oliver syndrome 5	AD		X									
NPC1	NM_000271	607623	257220	Niemann-Pick disease, type C1	AR			X								
NPC2	NM_006432	601015	607625	Niemann-pick disease, type C2	AR			X								
NPHP1	NM_000272	607100	609583	Joubert syndrome 4	AR			X								
NPHP3	NM_153240	608002	267010	Meckel syndrome 7	AR			X								
NPRL2	NM_006545	607072	617116	Epilepsy, familial focal, with variable foci 2	AD		X									
NPRL3	NM_001077350	600928	617118	Epilepsy, familial focal, with variable foci 3	AD		X									
NR2F1	NM_005654	132890	615722	Bosch-Boonstra-Schaaf optic atrophy syndrome	AD		X									
NR5A1	NM_004959	184757	612964	Adrenocortical insufficiency	AD		X									
NRAS	NM_002524	164790	613224	Noonan syndrome 6	AD		X									
NRXN1	NM_001135659	600565	614325	Pitt-Hopkins-like syndrome 2	AR			X								
NSD1	NM_022455	606681	117550	Sotos syndrome 1	AD		X									
NSDHL	NM_015922	300275	308050	CHILD syndrome	CI	X			X	X						
NSUN2	NM_017755	610916	611091	Mental retardation, autosomal recessive 5	AR			X								
NTNG2	NM_032536	618689	618718	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia	AR			X								
NUBPL	NM_025152	613621	618242	Mitochondrial complex I deficiency, nuclear type 21	AR			X								
OCLN	NM_002538	602876	251290	Pseudo-TORCH syndrome 1	AR			X								
OCRL	NM_000276	300535	309000	Lowe syndrome	XLR					X						
OFD1	NM_003611	300170	311200	Orofaciodigital syndrome I	CI	X			X	X						
OGT	NM_181672	300255	300997	Mental retardation, X-linked 106	XLR					X						

OPHN1	NM_002547	300127	300486	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	XLR					X						
ORC1	NM_004153	601902	224690	Meier-Gorlin syndrome 1	AR			X								
ORC4	NM_002552	603056	613800	Meier-Gorlin syndrome 2	AR			X								
ORC6	NM_014321	607213	613803	Meier-Gorlin syndrome 3	AR			X								
OSGEP	NM_017807	610107	617729	Galloway-Mowat syndrome 3	AR			X								
OTC	NM_000531	300461	311250	Ornithine transcarbamylase deficiency	XLR					X				X		
OTUD5	NM_017602	300713	301056	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked	XLR					X						
OTUD6B	NM_016023	612021	617452	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	AR			X								
OTX2	NM_172337	600037	610125	Microphthalmia, syndromic 5	AD		X									
P4HTM	NM_177938	614584	618493	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities	AR			X								
PACS1	NM_018026	607492	615009	Schuurs-Hoeijmakers syndrome	AD		X									
PACS2	NM_001100913	610423	618067	Developmental and epileptic encephalopathy 66	AD		X									
PAFAH1B1	NM_000430	601545	607432	Lissencephaly 1	AD		X									
PAH	NM_000277	612349	261600	Phenylketonuria	AR			X								
PAK3	NM_002578	300142	300558	Mental retardation, X-linked 30/47	XLR					X						
PANK2	NM_153638	606157	234200	Neurodegeneration with brain iron accumulation 1	AR			X								
PAX1	NM_006192	167411	615560	Otofaciocervical syndrome 2	AR			X								
PAX3	NM_181457	606597	122880	Craniofacial-deafness-hand syndrome	AD		X	-								
PAX6	NM_000280	607108	106210	Aniridia	AD		X									
PC	NM_000920	608786	266150	Pyruvate carboxylase deficiency	AR			X								
PCDH12	NM_016580	605622	251280	Diencephalic-mesencephalic junction dysplasia syndrome 1	AR			X								
PCDH19	NM_001184880	300460	300088	Epileptic encephalopathy, early infantile, 9	XLD				X					X		
PCGF2	NM_007144	600346	618371	Turnpenny-Fry syndrome	AD		X									
PCNT	NM_006031	605925	210720	Microcephalic osteodysplastic primordial dwarfism, type II	AR			X								
PCYT2	NM_001184917	602679	615033	Spastic paraplegia 82, autosomal recessive	AR			X								
PDE4D	NM_001104631	600129	614613	Acrodysostosis 2, with or without hormone resistance	AD		X									
PDE6D	NM_002601	602676	615665	Joubert syndrome 22	AR			X								
PDHA1	NM_000284	300502	312170	Pyruvate dehydrogenase E1-alpha deficiency	XLD				X					X		
PDHB	NM_000925	179060	614111	Pyruvate dehydrogenase E1-beta deficiency	AR			X								
PDHX	NM_003477	608769	245349	Lacticacidemia due to PDX1 deficiency	AR			X								
PDP1	NM_018444	605993	608782	Pyruvate dehydrogenase phosphatase deficiency	AR			X								
PDSS1	NM_014317	607429	614651	Coenzyme Q10 deficiency, primary, 2	AR			X								
PDSS2	NM_020381	610564	614652	Coenzyme Q10 deficiency, primary, 3	AR			X								
PEPD	NM_000285	613230	170100	Prolidase deficiency	AR			X								
PET100	NM_001171155	614770	619055	Mitochondrial complex IV deficiency, nuclear type 12	AR			X								
PEX1	NM_000466	602136	214100	Peroxisome biogenesis disorder 1A (Zellweger)	AR			X								
PEX10	NM_153818	602859	614870	Peroxisome biogenesis disorder 6A (Zellweger)	AR			X								
PEX11B	NM_003846	603867	614920	Peroxisome biogenesis disorder 14B	AR			X								
PEX12	NM_000286	601758	614859	Peroxisome biogenesis disorder 3A (Zellweger)	AR			X								
PEX13	NM_002618	601789	614883	Peroxisome biogenesis disorder 11A (Zellweger)	AR			X								
PEX14	NM_004565	601791	614887	Peroxisome biogenesis disorder 13A (Zellweger)	AR			X								
PEX16	NM_004813	603360	614876	Peroxisome biogenesis disorder 8A (Zellweger)	AR			X								
PEX19	NM_002857	600279	614886	Peroxisome biogenesis disorder 12A (Zellweger)	AR			X								
PEX2	NM_000318	170993	614866	Peroxisome biogenesis disorder 5A (Zellweger)	AR			X								
PEX26	NM_017929	608666	614872	Peroxisome biogenesis disorder 7A (Zellweger)	AR			X								
PEX3	NM_003630	603164	614882	Peroxisome biogenesis disorder 10A (Zellweger)	AR			X								
PEX5	NM_001131025	600414	214110	Peroxisome biogenesis disorder 2A (Zellweger)	AR			X								
PEX6	NM_000287	601498	614862	Peroxisome biogenesis disorder 4A (Zellweger)	AR			X								
PEX7	NM_000288	601757	614879	Peroxisome biogenesis disorder 9B	AR			X								
PGAP1	NM_024989	611655	615802	Mental retardation, autosomal recessive 42	AR			X								
PGAP2	NM_001256240	615187	614207	Hyperphosphatasia with mental retardation syndrome 3	AR			X								
PGAP3	NM_033419	611801	615716	Hyperphosphatasia with mental retardation syndrome 4	AR			X								
PGK1	NM_000291	311800	300653	Phosphoglycerate kinase 1 deficiency	XLR					X						
PGM1	NM_002633	171900	614921	Congenital disorder of glycosylation, type It	AR			X								
PHC1	NM_004426	602978	615414	Microcephaly 11, primary, autosomal recessive	AR			X								
PHF6	NM_032458	300414	301900	Borjeson-Forssman-Lehmann syndrome	XLR					X						
PHF8	NM_015107	300560	300263	Mental retardation syndrome, X-linked, Siderius type	XLR					X						
PHGDH	NM_006623	606879	601815	Phosphoglycerate dehydrogenase deficiency	AR			X								
PHIP	NM_017934	612870	617991	Chung-Jansen syndrome	AD		X									
PIBF1	NM_006346	607532	617767	Joubert syndrome 33	AR			X								
PIGA	NM_002641	311770	300868	Multiple congenital anomalies-hypotonia-seizures syndrome 2	XLR					X						
PIGB	NM_004855	604122	618580	Developmental and epileptic encephalopathy 80	AR			X								
PIGC	NM_002642	601730	601730	Glycosylphosphatidylinositol biosynthesis defect 16	AR			X								Edvardson et al., JMG 2017 : gl
PIGG	NM_001127178	616918	616917	Mental retardation, autosomal recessive 53	AR			X								
PIGL	NM_004278	605947	280000	CHIME syndrome	AR			X								
PIGN	NM_176787	606097	614080	Multiple congenital anomalies-hypotonia-seizures syndrome 1	AR			X								
PIGO	NM_032634	614730	614749	Hyperphosphatasia with mental retardation syndrome 2	AR			X								
PIGQ	NM_004204	605754	618548	Developmental and epileptic encephalopathy 77	AR			X								
PIGT	NM_015937	610272	615398	Multiple congenital anomalies-hypotonia-seizures syndrome 3	AR			X								
PIGV	NM_017837	610274	239300	Hyperphosphatasia with mental retardation syndrome 1	AR			X								
PIGW	NM_178517	610275	616025	Hyperphosphatasia with mental retardation syndrome 5	AR			X								
PIGY	NM_001042616	610662	616809	Hyperphosphatasia with mental retardation syndrome 6	AR			X								
PIK3R2	NM_005027	603157	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	AD		X							X		
PLA2G6	NM_003560	603604	610217	Neurodegeneration with brain iron accumulation 2B	AR			X								
PLCB1	NM_015192	607120	613722	Epileptic encephalopathy, early infantile, 12	AR			X								

PLCB4	NM_000933	600810	614669	Auriculocondylar syndrome 2	CI	X	X	X								
PLK4	NM_014264	605031	616171	Microcephaly and chorioretinopathy, autosomal recessive, 2	AR			X								
PLOD1	NM_000302	153454	225400	Ehlers-Danlos syndrome, type VI	AR			X								
PLP1	NM_000533	300401	312080	Pelizaeus-Merzbacher disease	XLR					X						
PLPBP	NM_007198	604436	617290	Epilepsy, early-onset, vitamin B6-dependent	AR			X								
PMM2	NM_000303	601785	212065	Congenital disorder of glycosylation, type Ia	AR			X								
PNKP	NM_007254	605610	613402	Microcephaly, seizures, and developmental delay	AR			X								
PNP	NM_000270	164050	613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency	AR			X								
PNPO	NM_018129	603287	610090	Pyridoxamine 5'-phosphate oxidase deficiency	AR			X								
PNPT1	NM_033109	610316	614932	Combined oxidative phosphorylation deficiency 13	AR			X								
POC1A	NM_015426	614783	614813	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	AR			X								
POGZ	NM_015100	614787	616364	White-Sutton syndrome	AD		X									
POLA1	NM_016937	312040	301030	Van Esch-O'Driscoll syndrome	XLR					X						
POLG	NM_002693	174763	613662	Mitochondrial DNA depletion syndrome 4B (MNGIE type)	CI	X	X	X								
POLH	NM_006502	603968	278750	Xeroderma pigmentosum, variant type	AR			X								
POLR2A	NM_000937	180660	618603	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities	AD		X									
POLR3A	NM_007055	614258	607694	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	AR			X								
POLR3B	NM_018082	614366	614381	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	AR			X								
POMGNT1	NM_017739	606822	613151	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	AR			X								
POMGNT2	NM_032806	614828	614830	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8	AR			X								
POMK	NM_032237	615247	615249	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	AR			X								
POMT1	NM_007171	607423	613155	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	AR			X								
POMT2	NM_013382	607439	613156	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	AR			X								
PORCN	NM_203475	300651	305600	Focal dermal hypoplasia	XLD				X							
POU3F3	NM_006236	602480	-	Snijders Blok et al., AJHG 2019 : developmental delays and/or intellectual disability and impairments in speech and language skills	AD		X									
PPM1D	NM_003620	605100	617450	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold	AD		X					X				
PPP1CB	NM_206876	600590	617506	Noonan syndrome-like disorder with loose anagen hair 2	AD		X									
PPP2R1A	NM_014225	605983	616362	Mental retardation, autosomal dominant 36	AD		X									
PPP2R5D	NM_006245	601646	616355	Mental retardation, autosomal dominant 35	AD		X									
PPT1	NM_000310	600722	256730	Ceroid lipofuscinosis, neuronal, 1	AR			X								
PQBP1	NM_005710	300463	309500	Renpenning syndrome	XLR					X						
PRICKLE1	NM_153026	608500	612437	Epilepsy, progressive myoclonic 1B	AR			X								
PRMT7	NM_019023	610087	617157	Short stature, brachydactyly, intellectual developmental disability, and seizures	AR			X								
PRPS1	NM_002764	311850	301835	Arts syndrome	XLR					X						
PRRT2	NM_145239	614386	605751	Seizures, benign familial infantile, 2	AD		X									
PRSS12	NM_003619	606709	249500	Mental retardation, autosomal recessive 1	AR			X								
PSAP	NM_002778	176801	611722	Krabbe disease, atypical	AR			X								
PSAT1	NM_058179	610936	616038	Neu-Laxova syndrome 2	AR			X								
PSMD12	NM_002816	604450	617516	Stankiewicz-Isidor syndrome	AD		X									
PTCH1	NM_000264	601309	610828	Holoprosencephaly 7	AD		X									
PTCHD1	NM_173495	300828	300830	Autism, susceptibility to, X-linked 4	XLR					X						
PTDSS1	NM_014754	612792	151050	Lenz-Majewski hyperostotic dwarfism	AD		X									
PTEN	NM_000314	601728	158350	Cowden syndrome 1	AD		X					X				
PTPN11	NM_002834	176876	151100	LEOPARD syndrome 1	AD		X									
PTRF	NM_012232	603198	613327	Lipodystrophy, congenital generalized, type 4	AR			X								
PTRH2	NM_016077	608625	616263	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	AR			X								
PTS	NM_000317	612719	261640	Hyperphenylalaninemia, BH4-deficient, A	AR			X								
PUF60	NM_078480	604819	615583	Verheij syndrome	AD		X									
PURA	NM_005859	600473	616158	Mental retardation, autosomal dominant 31	AD		X									
PUS1	NM_025215	608109	600462	Myopathy, lactic acidosis, and sideroblastic anemia 1	AR			X								
PYCR1	NM_006907	179035	612940	Cutis laxa, autosomal recessive, type IIB	AR			X								
QARS1	NM_005051	603727	615760	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	AR			X								
QDPR	NM_000320	612676	261630	Hyperphenylalaninemia, BH4-deficient, C	AR			X								
QRICH1	NM_017730	617387	617982	Ververi-Brady syndrome	AD		X									
RAB11B	NM_004218	604198	617807	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter	AD		X									
RAB18	NM_021252	602207	614222	Warburg micro syndrome 3	AR			X								
RAB23	NM_183227	606144	201000	Carpenter syndrome	AR			X								
RAB27A	NM_004580	603868	607624	Griscelli syndrome, type 2	AR			X								
RAB39B	NM_171998	300774	300271	Mental retardation, X-linked 72	XLR					X						
RAB3GAP1	NM_012233	602536	600118	Warburg micro syndrome 1	AR			X								
RAB3GAP2	NM_012414	609275	212720	Martsolf syndrome	AR			X								
RAC1	NM_018890	602048	617751	Mental retardation, autosomal dominant 48	AD		X									
RAD21	NM_006265	606462	614701	Cornelia de Lange syndrome 4	AD		X									
RAF1	NM_002880	164760	611553	Noonan syndrome 5	AD		X									
RAI1	NM_030665	607642	182290	Smith-Magenis syndrome	AD		X									
RARB	NM_000965	180220	615524	Microphthalmia, syndromic 12	AD		X	-								
RARS1	NM_002887	107820	616140	Leukodystrophy, hypomyelinating, 9	AR			X								
RARS2	NM_020320	611524	611523	Pontocerebellar hypoplasia, type 6	AR			X								
RASA2	NM_006506	601589	-	Aoki et al., JHG 2016 : RASopathie	AD		X					X				
RAX	NM_013435	601881	611038	Microphthalmia, isolated 3	AR			X								
RBBP8	NM_002894	604124	606744	Seckel syndrome 2	AR			X								

RBM10	NM_005676	300080	311900	TARP syndrome	XLR					X						
RBM28	NM_018077	612074	612079	Alopecia, neurologic defects, and endocrinopathy syndrome	AR			X								
RBM8A	NM_005105	605313	274000	Thrombocytopenia-absent radius syndrome	AR			X								
RBPJ	NM_005349	147183	614814	Adams-Oliver syndrome 3	AD		X									
RECQL4	NM_004260	603780	218600	Baller-Gerold syndrome	AR			X								
RELN	NM_005045	600514	257320	Lissencephaly 2 (Norman-Roberts type)	CI	X	X	X								
RERE	NM_012102	605226	616975	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	AD		X									
RFT1	NM_015138	611908	612015	Congenital disorder of glycosylation, type In	AR			X								
RHOBTB2	NM_001160036	607352	618004	Developmental and epileptic encephalopathy 64	AD		X									
RIT1	NM_006912	609591	615355	Noonan syndrome 8	AD		X									
RMND1	NM_017909	614917	614922	Combined oxidative phosphorylation deficiency 11	AR			X								
RNASEH2A	NM_006397	606034	610333	Aicardi-Goutieres syndrome 4	AR			X								
RNASEH2B	NM_024570	610326	610181	Aicardi-Goutieres syndrome 2	AR			X								
RNASEH2C	NM_032193	610330	610329	Aicardi-Goutieres syndrome 3	AR			X								
RNASET2	NM_003730	612944	612951	Leukoencephalopathy, cystic, without megalencephaly	AR			X								
RNF113A	NM_006978	300951	300953	Trichothiodystrophy 5, nonphotosensitive	XLD				X							
ROGDI	NM_024589	614574	226750	Kohlschutter-Tonz syndrome	AR			X								
RORB	NM_006914	601972	618357	{Epilepsy, idiopathic generalized, susceptibility to, 15}	AD		X									
RPGRIP1L	NM_015272	610937	611560	Joubert syndrome 7	AR			X								
RPIA	NM_144563	180430	608611	Ribose 5-phosphate isomerase deficiency	AR			X								
RPL10	NM_006013	312173	300998	Mental retardation, X-linked, syndromic, 35	XLR					X				X		
RPS6KA3	NM_004586	300075	303600	Coffin-Lowry syndrome	XLD				X							
RRAS	NM_006270	165090	-	Flex et al., HMG 2014 : RASopathie	AD		X					X				
RRAS2	NM_012250	600098	618624	Noonan syndrome 12	AD		X									
RRM2B	NM_015713	604712	612075	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	AR			X								
RSRC1	NM_016625	613352	618402	Intellectual developmental disorder, autosomal recessive 70	AR			X								
RTEL1	NM_032957	608833	615190	Dyskeratosis congenita, autosomal recessive 5	CI	X	X	X								
RTN4IP1	NM_032730	610502	616732	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	AR			X								
RTTN	NM_173630	610436	614833	Microcephaly, short stature, and polymicrogyria with seizures	AR			X								
RXYLT1	NM_014254	605862	615041	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	AR			X								
SALL1	NM_002968	602218	107480	Townes-Brocks branchiootorenal-like syndrome	AD		X									
SAMHD1	NM_015474	606754	612952	Aicardi-Goutieres syndrome 5	AR		-	X								
SATB1	NM_001195470	602075	619228	Developmental delay with dysmorphic facies and dental anomalies	AD		X									
SATB2	NM_015265	608148	612313	Glass syndrome	AD		X									
SC5D	NM_006918	602286	607330	Lathosterolosis	AR			X								
SCARB2	NM_005506	602257	254900	Epilepsy, progressive myoclonic 4, with or without renal failure	AR			X								
SCN1A	NM_001165963	182389	607208	Epileptic encephalopathy, early infantile, 6 (Dravet syndrome)	AD		X							X		
SCN1B	NM_001037	600235	604233	Epilepsy, generalized, with febrile seizures plus, type 1	CI	X	X	X								
SCN2A	NM_021007	182390	613721	Epileptic encephalopathy, early infantile, 11	AD		X							X		
SCN3A	NM_006922	182391	617938	Developmental and epileptic encephalopathy 62	AD		X									
SCN8A	NM_014191	600702	614558	Epileptic encephalopathy, early infantile, 13	AD		X									
SCO2	NM_005138	604272	604377	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	CI	X	X	X								
SDCCAG8	NM_006642	613524	615993	Bardet-Biedl syndrome 16	AR			X								
SDHAF1	NM_001042631	612848	252011	Mitochondrial complex II deficiency	AR			X								
SEPSECS	NM_016955	613009	613811	Pontocerebellar hypoplasia type 2D	AR			X								
SERAC1	NM_032861	614725	614739	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	AR			X								
SETBP1	NM_015559	611060	616078	Mental retardation, autosomal dominant 29	AD		X									
SETD2	NM_014159	612778	616831	Luscan-Lumish syndrome	AD		X									
SETD5	NM_001080517	615743	615761	Mental retardation, autosomal dominant 23	AD		X									
SGSH	NM_000199	605270	252900	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR			X								
SHANK2	NM_012309	603290	613436	Autism susceptibility 17	AD		X									
SHANK3	NM_001372044	606230	606232	Phelan-McDermid syndrome	AD		X									
SHH	NM_000193	600725	142945	Holoprosencephaly 3	AD		X									
SHOC2	NM_007373	602775	607721	Noonan-like syndrome with loose anagen hair	AD		X									
SHROOM4	NM_020717	300579	300434	Stocco dos Santos X-linked mental retardation syndrome	XLR					X						
SIK1	NM_173354	605705	616341	Developmental and epileptic encephalopathy 30	AD		X									
SIL1	NM_022464	608005	248800	Marinesco-Sjogren syndrome	AR			X								
SIN3A	NM_001145358	607776	613406	Witteveen-Kolk syndrome	AD		X									
SIX3	NM_005413	603714	157170	Holoprosencephaly 2	AD		X									
SKI	NM_003036	164780	182212	Shprintzen-Goldberg syndrome	AD		X									
SLC12A5	NM_020708	606726	616645	Developmental and epileptic encephalopathy 34	AR			X								
SLC12A6	NM_133647	604878	218000	Agenesis of the corpus callosum with peripheral neuropathy	AR			X								
SLC13A5	NM_177550	608305	615905	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta	AR			X								
SLC16A2	NM_006517	300095	300523	Allan-Herndon-Dudley syndrome	XLD				X							
SLC17A5	NM_012434	604322	604369	Salla disease	AR			X								
SLC19A3	NM_025243	606152	607483	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	AR			X								
SLC1A2	NM_004171	600300	617105	Developmental and epileptic encephalopathy 41	AD		X									
SLC1A4	NM_003038	600229	616657	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	AR			X								
SLC25A1	NM_005984	190315	615182	Combined D-2- and L-2-hydroxyglutaric aciduria	AR			X								
SLC25A12	NM_003705	603667	612949	Epileptic encephalopathy, early infantile, 39	AR			X								
SLC25A15	NM_014252	603861	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR			X								
SLC25A19	NM_021734	606521	607196	Microcephaly, Amish type	AR			X								
SLC25A22	NM_024698	609302	609304	Epileptic encephalopathy, early infantile, 3	AR			X								
SLC2A1	NM_006516	138140	606777	GLUT1 deficiency syndrome 1, infantile onset, severe	CI	X	X	X								

SLC33A1	NM_004733	603690	614482	Congenital cataracts, hearing loss, and neurodegeneration	AR		-	X								
SLC35A1	NM_006416	605634	603585	Congenital disorder of glycosylation, type IIf	AR			X								
SLC35A2	NM_001042498	314375	300896	Congenital disorder of glycosylation, type IIm	XLD				X							
SLC35A3	NM_012243	605632	615553	?Arthrogryposis, mental retardation, and seizures	AR			X								
SLC35C1	NM_018389	605881	266265	Congenital disorder of glycosylation, type IIc	AR			X								
SLC39A8	NM_022154	608732	616721	Congenital disorder of glycosylation, type IIn	AR			X								
SLC45A1	NM_001080397	605763	617532	Intellectual developmental disorder with neuropsychiatric features	AR			X								
SLC46A1	NM_080669	611672	229050	Folate malabsorption, hereditary	AR			X								
SLC4A4	NM_003759	603345	604278	Renal tubular acidosis, proximal, with ocular abnormalities	AR			X								
SLC6A1	NM_003042	137165	616421	Myoclonic-atonic epilepsy	AD		X									
SLC6A17	NM_001010898	610299	616269	Mental retardation, autosomal recessive 48	AR			X								
SLC6A3	NM_001044	126455	613135	Parkinsonism-dystonia, infantile	AR			X								
SLC6A5	NM_004211	604159	614618	Hyperekplexia 3	CI	X	X	X								
SLC6A8	NM_005629	300036	300352	Cerebral creatine deficiency syndrome 1	XLR					X					X	
SLC7A7	NM_001126106	603593	222700	Lysinuric protein intolerance	AR			X								
SLC9A6	NM_006359	300231	300243	Mental retardation, X-linked syndromic, Christianson type	XLD				X							
SMARCA2	NM_003070	600014	601358	Nicolaides-Baraitser syndrome	AD		X									
SMARCA4	NM_001128849	603254	614609	Coffin-Siris syndrome 4	AD		X									
SMARCB1	NM_003073	601607	614608	Coffin-Siris syndrome 3	AD		X						X			
SMARCE1	NM_003079	603111	616938	Coffin-Siris syndrome 5	AD		X									
SMC1A	NM_006306	300040	300590	Cornelia de Lange syndrome 2	XLD				X							
SMC3	NM_005445	606062	610759	Cornelia de Lange syndrome 3	AD		X									
SMOC1	NM_001034852	608488	206920	Microphthalmia with limb anomalies	AR			X							X	
SMPD1	NM_000543	607608	257200	Niemann-Pick disease, type A	AR			X								
SMS	NM_004595	300105	309583	Mental retardation, X-linked, Snyder-Robinson type	XLR					X						
SNAP25	NM_003081	600322	-	Klöckner et al., Genet Med 2021 : early-onset developmental and epileptic encephalopathy	AD		X									
SNAP29	NM_004782	604202	609528	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR			X								
SNIP1	NM_024700	608241	614501	Psychomotor retardation, epilepsy, and craniofacial dysmorphism	AR			X								
SNX14	NM_153816	616105	616354	Spinocerebellar ataxia, autosomal recessive 20	AR			X								
SOBP	NM_018013	613667	613671	Mental retardation, anterior maxillary protrusion, and strabismus	AR			X								
SON	NM_032195	182465	617140	ZTTK syndrome	AD		X									
SOS1	NM_005633	182530	610733	Noonan syndrome 4	AD		X									
SOS2	NM_006939	601247	616559	Noonan syndrome 9	AD		X									
SOX10	NM_006941	602229	611584	Waardenburg syndrome, type 2E, with or without neurologic involvement	AD		X									
SOX11	NM_003108	600898	615866	Mental retardation, autosomal dominant, 27	AD		X									
SOX2	NM_003106	184429	206900	Optic nerve hypoplasia and abnormalities of the central nervous system	AD		X									
SOX5	NM_006940	604975	616803	Lamb-Shaffer syndrome	AD		X									
SPATA5	NM_145207	613940	616577	Epilepsy, hearing loss, and mental retardation syndrome	AR			X								
SPECC1L	NM_015330	614140	145410	Opitz GBBB syndrome, type II	AD		X									
SPR	NM_003124	182125	612716	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	CI	X	X	X								
SPRED1	NM_152594	609291	611431	Legius syndrome	AD		X									
SPTAN1	NM_001130438	182810	613477	Epileptic encephalopathy, early infantile, 5	AD		X									
SPTBN4	NM_020971	606214	617519	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	AR			X								
SRCAP	NM_006662	611421	136140	Floating-Harbor syndrome	AD		X									
SRD5A3	NM_024592	611715	612379	Congenital disorder of glycosylation, type Iq	AR			X								
ST3GAL3	NM_006279	606494	611090	Mental retardation, autosomal recessive 12	AR			X								
ST3GAL5	NM_003896	604402	609056	Salt and pepper developmental regression syndrome	AR			X								
STAG1	NM_005862	604358	617635	Mental retardation, autosomal dominant 47	AD		X									
STAG2	NM_001042749	300826	301043	Holoprosencephaly 13, X-linked	XLR					X						
STAMBP	NM_006463	606247	614261	Microcephaly-capillary malformation syndrome	AR			X								
STIL	NM_003035	181590	612703	Microcephaly 7, primary, autosomal recessive	AR			X								
STRA6	NM_022369	610745	601186	Microphthalmia, syndromic 9	AR			X								
STRADA	NM_001003787	608626	611087	Polyhydramnios, megalencephaly, and symptomatic epilepsy	AR			X								
STT3A	NM_001278503	601134	615596	Congenital disorder of glycosylation, type Iw	AR			X								
STT3B	NM_178862	608605	615597	Congenital disorder of glycosylation, type Ix	AR			X								
STX1B	NM_052874	601485	616172	Generalized epilepsy with febrile seizures plus, type 9	AD		X									
STXBP1	NM_003165	602926	612164	Epileptic encephalopathy, early infantile, 4	AD		X									
SUCLA2	NM_003850	603921	612073	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	AR			X								
SUCLG1	NM_003849	611224	245400	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	AR			X								
SUMF1	NM_182760	607939	272200	Multiple sulfatase deficiency	AR			X								
SUOX	NM_000456	606887	272300	Sulfite oxidase deficiency	AR			X								
SURF1	NM_003172	185620	256000	Leigh syndrome, due to COX IV deficiency	AR			X						X		
SUZ12	NM_015355	606245	618786	Imagawa-Matsumoto syndrome	AD		X									
SVBP	NM_199342	617853	-	Igbal et al., Genet Med 2019	AR			X								
SYN1	NM_133499	313440	300491	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	CI	X			X	X						
SYNCRIP	NM_001159673	616686	-	Lelieveld et al., Nat Neurosci 2016 : ID	AD		X									
SYNE1	NM_033071	608441	610743	Spinocerebellar ataxia, autosomal recessive 8	CI	X	X	X								
SYNGAP1	NM_006772	603384	612621	Mental retardation, autosomal dominant 5	AD		X									
SYNJ1	NM_003895	604297	617389	Developmental and epileptic encephalopathy 53	AR			X								
SYP	NM_003179	313475	300802	Mental retardation, X-linked 96	XLR					X						
SYT14	NM_001146261	610949	614229	Spinocerebellar ataxia, autosomal recessive 11	AR			X								
SZT2	NM_015284	615463	615476	Epileptic encephalopathy, early infantile, 18	AR			X								
TACO1	NM_016360	612958	619052	Mitochondrial complex IV deficiency, nuclear type 8	AR			X								
TAF1	NM_004606	313650	300966	Mental retardation, X-linked, syndromic 33	XLR					X						
TAF2	NM_003184	604912	615599	Mental retardation, autosomal recessive 40	AR			X								

TAOK1	NM_020791	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.	AD			X									
TBC1D20	NM_144628	611663	615663	Warburg micro syndrome 4	AR				X								
TBC1D23	NM_001199198	617687	617695	Pontocerebellar hypoplasia, type 11	AR				X								
TBC1D24	NM_001199107	613577	615338	Epileptic encephalopathy, early infantile, 16	AR				X								
TBC1D7	NM_001143965	612655	248000	Macrocephaly/megalencephaly syndrome, autosomal recessive	AR				X								
TBCD	NM_005993	604649	617193	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	AR				X								
TBCE	NM_003193	604934	241410	Hypoparathyroidism-retardation-dysmorphism syndrome	AR				X								
TBCK	NM_001163435	616899	616900	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	AR				X								
TBL1XR1	NM_024665	608628	616944	Mental retardation, autosomal dominant 41	AD			X									
TBR1	NM_006593	604616	606053	Intellectual developmental disorder with autism and speech delay	AD			X									
TCF20	NM_005650	603107	618430	Developmental delay with variable intellectual impairment and behavioral abnormalities	AD			X									
TCF4	NM_001083962	602272	610954	Pitt-Hopkins syndrome	AD			X									
TCOF1	NM_001135243	606847	154500	Treacher Collins syndrome 1	AD			X									
TCTN1	NM_001082538	609863	614173	Joubert syndrome 13	AR				X								
TCTN2	NM_024809	613846	616654	Joubert syndrome 24	AR				X								
TCTN3	NM_015631	613847	614815	Joubert syndrome 18	AR				X								
TECPR2	NM_014844	615000	615031	Spastic paraplegia 49, autosomal recessive	AR				X								
TECR	NM_138501	610057	614020	Mental retardation, autosomal recessive 14	AR				X								
TFAP2A	NM_001042425	107580	113620	Branchiooculofacial syndrome	AD			X									
TGIF1	NM_173208	602630	142946	Holoprosencephaly 4	AD			X									
TH	NM_199292	191290	605407	Segawa syndrome, recessive	AR				X								
THOC2	NM_001081550	300395	300957	Mental retardation, X-linked 12/35	XLR					X							
THOC6	NM_024339	615403	613680	Beaulieu-Boycott-Innes syndrome	AR				X								
THRA	NM_199334	190120	614450	Hypothyroidism, congenital, nongoitrous, 6	AD			X									
TIMM8A	NM_004085	300356	304700	Mohr-Tranebjaerg syndrome	XLR					X							
TINF2	NM_001099274	604319	613990	Dyskeratosis congenita, autosomal dominant 3	AD			X									
TLK2	NM_006852	608439	618050	Mental retardation, autosomal dominant 57	AD			X									
TMCO1	NM_019026	614123	213980	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	AR				X								
TMEM107	NM_032354	616183	617563	Orofaciodigital syndrome XVI	AR				X								
TMEM138	NM_016464	614459	614465	Joubert syndrome 16	AR				X								
TMEM165	NM_018475	614726	614727	Congenital disorder of glycosylation, type IIk	AR				X								
TMEM216	NM_001173990	613277	608091	Joubert syndrome 2	AR				X								
TMEM231	NM_001077416	614949	614970	Joubert syndrome 20	AR				X								
TMEM237	NM_001044385	614423	614424	Joubert syndrome 14	AR				X								
TMEM67	NM_153704	609884	610688	Joubert syndrome 6	AR				X								
TMEM70	NM_017866	612418	614052	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	AR				X								
TNK2	NM_001010938	606994	-	Hitomi et al., Ann Neurol 2013 : severe autosomal recessive infantile-onset epilepsy and ID	AR				X								
TOGARAM1	NM_015091	617618	619185	Joubert syndrome 37	AR				X								
TPK1	NM_022445	606370	614458	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	AR				X								
TPP1	NM_000391	607998	609270	Spinocerebellar ataxia, autosomal recessive 7	AR				X								
TPP2	NM_003291	190470	619220	Immunodeficiency 78 with autoimmunity and developmental delay	AR				X								
TRAPPC11	NM_021942	614138	615356	Muscular dystrophy, limb-girdle, type 2S	AR				X								
TRAPPC4	NM_016146	610971	618741	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	AR				X								
TRAPPC9	NM_031466	611966	613192	Mental retardation, autosomal recessive 13	AR				X								
TREX1	NM_033629	606609	225750	Aicardi-Goutieres syndrome 1, dominant and recessive	CI	X		X									
TRIM32	NM_012210	602290	615988	Bardet-Biedl syndrome 11	AR				X								
TRIM8	NM_030912	606125	-	McClatchey et al., EJHG 2020 : Focal segmental glomerulosclerosis and mild intellectual disability	AD			X									
TRIO	NM_007118	601893	617061	Mental retardation, autosomal dominant 44	AD			X									
TRIP12	NM_004238	604506	617752	Mental retardation, autosomal dominant 49	AD			X									
TRMT10A	NM_152292	616013	616033	Microcephaly, short stature, and impaired glucose metabolism 1	AR				X								
TRMU	NM_018006	610230	613070	Liver failure, transient infantile	AR				X								
TRPM6	NM_017662	607009	602014	Hypomagnesemia 1, intestinal	AR				X								
TRPV4	NM_021625	605427	184252	Spondylometaphyseal dysplasia, Kozlowski type	AD			X									
TRRAP	NM_003496	603015	618454	Developmental delay with or without dysmorphic facies and autism	AD			X									
TSEN2	NM_025265	608753	612389	Pontocerebellar hypoplasia type 2B	AR				X								
TSEN54	NM_207346	608755	277470	Pontocerebellar hypoplasia type 2A	AR				X								
TSFM	NM_001172696	604723	610505	Combined oxidative phosphorylation deficiency 3	AR				X								
TSPAN7	NM_004615	300096	300210	Mental retardation, X-linked 58	XLR					X							
TTC19	NM_017775	613814	615157	Mitochondrial complex III deficiency, nuclear type 2	AR				X								
TTC37	NM_014639	614589	222470	Trichohepatoenteric syndrome 1	AR				X								
TTC8	NM_198309	608132	615985	Bardet-Biedl syndrome 8	AR				X								
TTI2	NM_001102401	614426	615541	Mental retardation, autosomal recessive 39	AR				X								
TUBA1A	NM_006009	602529	611603	Lissencephaly 3	AD			X									
TUBA8	NM_018943	605742	613180	Cortical dysplasia, complex, with other brain malformations 8	AR				X								
TUBB	NM_178014	191130	615771	Cortical dysplasia, complex, with other brain malformations 6	AD			X									
TUBB2A	NM_001069	615101	615763	Cortical dysplasia, complex, with other brain malformations 5	AD			X									
TUBB2B	NM_178012	612850	610031	Cortical dysplasia, complex, with other brain malformations 7	AD			X									
TUBB3	NM_006086	602661	614039	Cortical dysplasia, complex, with other brain malformations 1	AD			X									
TUBB4A	NM_006087	602662	612438	Leukodystrophy, hypomyelinating, 6	AD			X									
TUBG1	NM_001070	191135	615412	Cortical dysplasia, complex, with other brain malformations 4	AD			X									
TUBGCP6	NM_020461	610053	251270	Microcephaly and chorioretinopathy, autosomal recessive, 1	AR				X								
TUSC3	NM_006765	601385	611093	Mental retardation, autosomal recessive 7	AR				X								
TWIST1	NM_000474	601622	101400	Saethre-Chotzen syndrome	AD			X									

TWNK	NM_021830	606075	609286	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3	CI	X	X	X									
UBA5	NM_024818	610552	617132	Developmental and epileptic encephalopathy 44	AR			X									
UBE2A	NM_003336	312180	300860	Mental retardation, X-linked syndromic, Nascimento-type	XLR					X							
UBE3A	NM_130838	601623	105830	Angelman syndrome	AD		X									X	
UBE3B	NM_130466	608047	244450	Kaufman oculocerebrofacial syndrome	AR			X									
UBR1	NM_174916	605981	243800	Johanson-Blizzard syndrome	AR			X									
UFC1	NM_016406	610554	618076	Neurodevelopmental disorder with spasticity and poor growth	AR			X									
UFM1	NM_001286704	610553	617899	Leukodystrophy, hypomyelinating, 14	AR			X									
UNC80	NM_032504	612636	616801	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	AR			X									
UPF3B	NM_080632	300298	300676	Mental retardation, X-linked, syndromic 14	XLR					X							
UQCRQ	NM_014402	612080	615159	Mitochondrial complex III deficiency, nuclear type 4	AR			X									
UROC1	NM_144639	613012	276880	Urocanase deficiency	AR			X									
USP18	NM_017414	607057	617397	Pseudo-TORCH syndrome 2	AR			X									
USP9X	NM_001039590	300072	300919	Mental retardation, X-linked 99	CI	X			X	X							
VARS2	NM_001167734	612802	615917	Combined oxidative phosphorylation deficiency 20	AR			X									
VLDLR	NM_003383	192977	224050	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	AR			X									
VPS13B	NM_017890	607817	216550	Cohen syndrome	AR			X									
VPS4A	NM_013245	609982	619273	CIMDAG syndrome	AD		X										
VPS53	NM_001128159	615850	615851	Pontocerebellar hypoplasia, type 2E	AR			X									
VRK1	NM_003384	602168	607596	Pontocerebellar hypoplasia type 1A	AR			X									
WAC	NM_016628	615049	616708	Desanto-Shinawi syndrome	AD		X										
WDPCP	NM_015910	613580	615992	Bardet-Biedl syndrome 15	AR			X									
WDR26	NM_025160	617424	617616	Skraban-Deardorff syndrome	AD		X										
WDR37	NM_014023	618586	618652	Neurooculocardiogenitourinary syndrome	AD		X										
WDR45	NM_007075	300526	300894	Neurodegeneration with brain iron accumulation 5	XLD				X								
WDR45B	NM_019613	609226	617977	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	AR			X									
WDR62	NM_001083961	613583	604317	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	AR			X									
WDR73	NM_032856	616144	251300	Galloway-Mowat syndrome	AR			X									
WDR81	NM_001163809	614218	610185	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	AR			X									
WWOX	NM_016373	605131	616211	Epileptic encephalopathy, early infantile, 28	AR			X									
XPA	NM_000380	611153	278700	Xeroderma pigmentosum, group A	AR			X									
XPC	NM_004628	613208	278720	Xeroderma pigmentosum, group C	AR			X									
XYLT1	NM_022166	608124	615777	Desbuquois dysplasia 2	AR			X									
YAP1	NM_001130145	606608	120433	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation	AD		X										
YWHAG	NM_012479	605356	617665	Developmental and epileptic encephalopathy 56	AD		X										
YY1	NM_003403	600013	617557	Gabriele-de Vries syndrome	AD		X										
ZBTB16	NM_006006	176797	612447	Skeletal defects, genital hypoplasia, and mental retardation	AR			X									
ZBTB18	NM_205768	608433	612337	Mental retardation, autosomal dominant 22	AD		X										
ZBTB20	NM_001164342	606025	259050	Primrose syndrome	AD		X										
ZBTB24	NM_014797	614064	614069	Immunodeficiency-centromeric instability-facial anomalies syndrome-2	AR			X									
ZC4H2	NM_018684	300897	314580	Wieacker-Wolff syndrome	XLR					X							
ZDHHC9	NM_016032	300646	300799	Mental retardation, X-linked syndromic, Raymond type	XLR					X							
ZEB2	NM_014795	605802	235730	Mowat-Wilson syndrome	AD		X										
ZFYVE26	NM_015346	612012	270700	Spastic paraplegia 15, autosomal recessive	AR			X									
ZIC1	NM_003412	600470	618736	Structural brain anomalies with impaired intellectual development and craniosynostosis	AD		X										
ZIC2	NM_007129	603073	609637	Holoprosencephaly 5	AD		X										
ZMPSTE24	NM_005857	606480	608612	Mandibuloacral dysplasia with type B lipodystrophy	AR			X									
ZMYND11	NM_006624	608668	616083	Mental retardation, autosomal dominant 30	AD		X										
ZNF335	NM_022095	610827	615095	Microcephaly 10, primary, autosomal recessive	AR			X									
ZNF423	NM_015069	604557	614844	Joubert syndrome 19	CI	X	X	X									
ZNF711	NM_021998	314990	300803	Mental retardation, X-linked 97	XLR					X							
ZSWIM6	NM_020928	615951	617865	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features	AD		X										

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

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CABERG Jean-Hubert le 28/04/2023