Samenstelling van het ID_Gene_Panelv6 genenpanel

gen	NCBI gen id	overerving	Geassocieerd syndroom, OMIM-nr
AAAS	8086	AR	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	16	AD/AR	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AASS	10157	AR	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	18	AR	GABA-transaminase deficiency, 613163
ABCC9	10060	AD	Atrial fibrillation, familial, 12, 614050|Cardiomyopathy, dilated, 1O, 608569|Hypertrichotic osteochondrodysplasia, 239850
ABCD1	215	XLR	Adrenoleukodystrophy, 300100|Adrenomyeloneuropathy, adult, 300100
ABCD4	5826	AR	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	51099	AR	Chanarin-Dorfman syndrome, 275630
ACAD9	28976	AR	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADS	35	AR	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACAT1	38	AR	Alpha-methylacetoacetic aciduria, 203750
ACO2	50	AR	Infantile cerebellar-retinal degeneration, 614559
ACOX1	51	AR	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	197322	AR	Combined malonic and methylmalonic aciduria, 614265
ACSL4	2182	XL	Mental retardation, X-linked 63, 300387
ACTB	60	AD	Baraitser-Winter syndrome 1, 243310
ACTG1	71	AD	Baraitser-Winter syndrome 2, 614583
ACTL6A	86	AD	developmental delay; intellectual disability (GenomicsEngland,
ACTL6B	51412	AR	Karaca et al neuron, 2015
ACVR1	90	AD	Fibrodysplasia ossificans progressiva, 135100
ACY1	95	AR	Aminoacylase 1 deficiency, 609924
ADAM22	53616	AD	PMID:27066583
ADAMTS9	56999	AR	PMID:30609407
ADAR	103	AR	Aicardi-Goutieres syndrome 6, 615010|Dyschromatosis symmetrica hereditaria, 127400
ADARB1	104	AR	PMID:32719099
ADAT3	113179	AR	Mental retardation, autosomal recessive 36, 615286
ADGRG1	9289	AR	AR. . GPR56 Polymicrogyria bilateral frontoparietal 66854
ADGRG6	57211	AR	PMID: 30549416 (GPR126)
ADK	132	AR	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	23394	AD	Helsmoortel-van der Aa syndrome, 615873
ADPRHL2	54936	AR	PMID: 30401461
ADRA2B	151	AD/AR	AUTOSOMAL RECESSIVE MENTAL RETARDATION, OMIM:607876(Intellectual disability)
ADSL	158	AR	Adenylosuccinase deficiency, 103050
AFF2	2334	XLR	Mental retardation, X-linked, FRAXE type, 309548
AFF4	27125	AD	CHOPS syndrome, 616368
AFG3L2	10939	AR	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	175	AR	Aspartylglucosaminuria, 208400
AGO1	26523	AD	PMID: 30213762
AGO2	27161	AD	PMID:33199684
AGTR2	186		Mental retardation, X-linked (Ylisaukko-oja (2004) Hum Genet 114, 211)|?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476)|?Mental retardation, pervasive developmental disorder and epilepsy (Takeshita (2012) Brain Dev epub,
AHCY	191	AR	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	27245	AD	Xia-Gibbs syndrome, 615829

AHI1	54806	AR	Joubert syndrome-3, 608629
AIFM1	9131	XLR	Combined oxidative phosphorylation deficiency 6, 300816|Cowchock syndrome, 310490|Deafness, X-linked 5, 300614
AIMP1	9255	AR	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	7965	AR	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	10000	AD	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	5832	AD/AR	Cutis laxa, autosomal dominant 3, 616603|Cutis laxa, autosomal recessive, type IIIA, 219150|Spastic paraplegia 9A, autosomal dominant, 601162|Spastic paraplegia 9B, autosomal recessive, 616586
ALDH3A2	224	AR	Sjogren-Larsson syndrome, 270200
ALDH4A1	8659	AR	Hyperprolinemia, type II, 239510
ALDH5A1	7915	AR	Succinic semialdehyde dehydrogenase deficiency, 271980|pubmed 32621952
ALDH7A1	501	AR	Epilepsy, pyridoxine-dependent, 266100
ALG1	56052	AR	Congenital disorder of glycosylation, type Ik, 608540
ALG11	440138	AR	Congenital disorder of glycosylation, type Ip, 613661
ALG12	79087	AR	Congenital disorder of glycosylation, type Ig, 607143
ALG13	79868	XLR	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	85365	AR	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	10195	AR	Congenital disorder of glycosylation, type Id, 601110
ALG6	29929	AR	Congenital disorder of glycosylation, type Ic, 603147
ALG8	79053		Congenital disorder of glycosylation, type Ih, 608104
ALG9	79796	AR	Congenital disorder of glycosylation, type Il, 608776|Gillessen-Kaesbach-Nishimura syndrome, 263210
ALKBH8	91801	AR	PMID:31079898, Intellectual developmental disorder, autosomal recessive 71 618504 AR
ALMS1	7840	AR	Alstrom syndrome, 203800
ALX4	60529	AD/AR	Frontonasal dysplasia 2, 613451|Parietal foramina 2, 609597|{Craniosynostosis 5, susceptibility to}, 615529
AMER1	139285	XLD	OMIM300373
AMPD2	271	AR	Pontocerebellar hypoplasia, type 9, 615809
AMT	275	AR	Glycine encephalopathy, 605899
ANK2	287	AD	Autism Spectrum Disorders,phil et al Ped neurology 2017| OMIM:600919: Long QT syndrome 4; Cardiac arrhythmia, ankyrin-B-related
ANK3	288	AR	?Mental retardation, autosomal recessive, 37, 615493
ANKH	56172	AD	Chondrocalcinosis 2, 118600|Craniometaphyseal dysplasia, 123000
ANKRD11	29123	AD	KBG syndrome, 148050
ANO10	55129	AR	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	84168	AR	GAPO syndrome, 230740
AP1B1	162	AR	PMID:31630791
AP1S1	1174	AR	MEDNIK syndrome, 609313
AP1S2	8905	XL	Mental retardation, X-linked syndromic 5, 304340
AP2M1	1173	AD	PMID:31104773
AP3B1	8546	AR	Hermansky-Pudlak syndrome 2, 608233
AP3B2	8120	AR	Epileptic encephalopathy, early infantile, 48
AP4B1	10717	AR	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	23431	AR	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	9179	AR	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	11154	AR	Spastic paraplegia 52, autosomal recessive, 614067
APC2	10297	AR	PMID:31585108
APTX	54840	AR	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	372	AD	Short stature, rhizomelic, with microcephaly, micrognathia, and

			developmental delay, 617164
ARFGEF2	10564	AR	Periventricular heterotopia with microcephaly, 608097
ARG1	383	AR	Argininemia, 207800
ARHGAP31	57514	AD	Adams-Oliver syndrome 1 100300 AD
ARHGEF6	9459	XLR	Mental retardation, X-linked 46, 300436
ARHGEF9	23229	XLR	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	8289	AD	Coffin-Siris syndrome 2, 614607
ARID1B	57492	AD	Coffin-Siris syndrome 1, 135900
ARID2	196528	AD	Coffin-Siris syndrome 6, 617808
ARL13B	200894	AR	Joubert syndrome 8, 612291
ARL3	403	AR	PMID: 30269812
ARL6	84100	AR	Bardet-Biedl syndrome 3, 600151
ARMC9	80210	AR	Joubert syndrome; Intellectual Disability1 617622
ARSA	410	AR	Metachromatic leukodystrophy, 250100
ARSL	415	XLR	Chondrodysplasia punctata, X-linked recessive, 302950 (ARSE)
ARV1	64801	AR	Epileptic encephalopathy, early infantile, 38, 617020
ARX	170302	XLR	Epileptic encephalopathy, early infantile, 1, 308350|Hydranencephaly with abnormal genitalia, 300215|Lissencephaly, X-linked 2, 300215|Mental retardation, X-linked 29 and others, 300419
ASAH1	427	AR	Farber lipogranulomatosis, 228000
ASH1L	55870	AD	Autism (GenomicsEngland,
ASL	435	AR	Argininosuccinic aciduria, 207900
ASNS	440	AR	Asparagine synthetase deficiency, 615574
ASPA	443	AR	Canavan disease, 271900
ASPM	259266	AR	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	445	AR	Citrullinemia, 215700
ASXL1	171023	AD	Bohring-Opitz syndrome, 605039
ASXL2	55252	AD	Shashi-Pena syndrome, 617190
ASXL3	80816	AD	Bainbridge-Ropers syndrome, 615485
ATAD3A	55210	AR	AD/AR.Harel-Yoon syndrome 617183
ATIC	471	AR	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	51062	AD	Neuropathy, hereditary sensory, type ID, 613708
ATM	472	AR	ATAXIA-TELANGIECTASIA; AT, OMIM:208900(Choreoathetosis)
ATN1	1822	AD	PMID:30827498
ATP1A1	476	AD	PMID: 30388404
ATP1A2	477	AD	Alternating hemiplegia of childhood, 104290|Migraine, familial basilar, 602481|Migraine, familial hemiplegic, 2, 602481
ATP1A3	478	AD	AD.RAPID-ONSET DYSTONIA-PARKINSONISM OMIM614820
ATP2A2	488	AD	Acrokeratosis verruciformis, 101900|Darier disease, 124200
ATP6AP2	10159	AR	?Parkinsonism with spasticity, X-linked, 300911|?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	23545	AR	Cutis laxa, autosomal recessive, type IIA, 219200|Wrinkly skin syndrome, 278250
ATP6V0C	527	AD	PMID:33190975
ATP6V1A	523	AD	PMID:26350515
ATP6V1B2	526	AD	PMID: 31655144
ATP7A	538	XLR	Menkes disease, 309400
ATP8A2	51761	AR	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATR	545	AR	Seckel syndrome 1, 210600
ATRX	546	XLD/XLR	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448|Alpha-thalassemia/mental retardation syndrome, 301040
AUH	549	AR	3-methylglutaconic aciduria, type I, 250950
AUTS2	26053	AD	Mental retardation, autosomal dominant 26,615834
AVPR2	554	XLR	Diabetes insipidus, nephrogenic, 304800|Nephrogenic syndrome of inappropriate antidiuresis, 300539

B3GALNT2	148789	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	126792	AR	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349
B3GLCT	145173	AR	AR.Peters-plus syndrome 26154
B4GALNT1	2583	AR	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	2683	AR	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	11285	AR	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BAZ2B	29994	AD	PMID:31999386
BBS1	582	AR	Bardet-Biedl syndrome 1, 209900
BBS10	79738	AR	Bardet-Biedl syndrome 10, 615987
BBS12	166379	AR	Bardet-Biedl syndrome 12, 615989
BBS2	583	AR	Bardet-Biedl syndrome 2, 615981
BBS4	585	AR	Bardet-Biedl syndrome 4, 615982
BBS5	129880	AR	Bardet-Biedl syndrome 5, 615983
BBS7	55212	AR	Bardet-Biedl syndrome 7, 615984
BBS9	27241	AR	Bardet-Biedl syndrome 9, 615986
BCAP31	10134	XLR	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	593	AR	Maple syrup urine disease, type Ia, 248600
BCKDHB	594	AR	Maple syrup urine disease, type Ib, 248600
BCKDK	10295	AR	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL11A	53335	AD	Intellectual development disorder with persistence of fetal hemoglobin, 617101
BCL11B	64919	AD	PMID: 31347296; OMIM #618092
BCOR	54880	XL	Microphthalmia, syndromic 2, 300166
BCORL1	63035	XR	PMID:30941876
BCS1L	617	Mitochondrial/AR	Bjornstad syndrome, 262000|GRACILE syndrome, 603358|Leigh syndrome, 256000
BICD2	23299	AD	PMID: 27751653
BICRA	29998	?	PMID:33232675
BLM	641	AR	Bloom syndrome, 210900
BMP4	652	AD	OMIM607932
BOLA3	388962	AR	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	2186	AD	AD.Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. #617755
BRAF	673	AD	Adenocarcinoma of lung, somatic, 211980|LEOPARD syndrome 3, 613707|Noonan syndrome 7, 613706
BRAT1	221927	AR	PMID: 25319849
BRF1	2972	AR	PMID:25834187
BRPF1	7862	AD	AD.Intellectual developmental disorder with dysmorphic facies and ptosis 617333 OMIM617333 Intellectual developmental disorder with dysmorphic facies and ptosis 617333
BRSK2	9024	AD	PMID:30879638
BRWD3	254065	XLR	Mental retardation, X-linked 93, 300659|pubmled 31714006 (BRDW)
BSCL2	26580	AD/AR	Encephalopathy, progressive, with or without lipodystrophy, 615924|Neuropathy, distal hereditary motor, type VA, 600794
BTD	686	AR	Biotinidase deficiency, 253260
BUB1B	701	AR	Colorectal cancer, somatic, 114500
C12ORF4	57102	AR	Mental retardation, autosomal recessive 66, 618221
C12ORF57	113246	AR	Temtamy syndrome, 218340
C12ORF65	91574	AR	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C2CD3	26005	AR	Orofaciodigital syndrome XIV, 615948
CA2	760	AR	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA8	767	AR	Cerebellar ataxia and mental retardation with or without

			quadrupedal locomotion 3, 613227
CACNA1A	773	AD	Episodic ataxia, type 2, 108500
CACNA1B	774	AR	PMID:30982612
CACNA1C	775	AD	Timothy syndrome 601005 AD
CACNA1D	776	AD	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	777	AD	PMID: 30343943
CACNA1G	8913	AD/AR	AUTOSOMAL RECESSIVE MENTAL RETARDATION, OMIM:616795(Spastic ataxia|Cerebellar atrophy)
CACNA2D2	9254	AR	OMIM618501
CAD	790	AD	Epileptic encephalopathy, early infantile, 50, 616457
CAMK2A	815	AD	Mental retardation, autosomal dominant 53, 617798
CAMK2B	816	AD	AD.Mental retardation autosomal dominant 54 #617799
CAMTA1	23261	AD	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN10	11132	AR	PMID: 25773692
CAPZA2	830	AD	PMID:32338762
CARS1	833	AR	PMID:30824121|alt name CARS
CARS2	79587	AR	Combined oxidative phosphorylation deficiency 27, 616672
CASK	8573	XLD	FG syndrome 4, 300422|Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749|Mental retardation, with or without nystagmus, 300422
CBL	867	AD	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	875	AR	Homocystinuria, B6-responsive and nonresponsive types, 236200
CC2D1A	54862	AR	Mental retardation, autosomal recessive 3, 608443
CC2D2A	57545	AR	COACH syndrome, 216360|Joubert syndrome 9, 612285|Meckel syndrome 6, 612284
CCBE1	147372	AR	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC115	84317	AR	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	51244	AR	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	28952	XLR	Ritscher-Schinzel syndrome 2, 300963
CCDC47	57003	AR	PMID: 30401460
CCDC78	124093	AD	Myopathy, centronuclear, 4, 614807
CCDC88A	55704	AR	?PEHO syndrome-like, 617507
CCDC88C	440193	AD/AR	?Spinocerebellar ataxia 40, 616053; Hydrocephalus, congenital, 1 236600
CCND2	894	AD	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3 615938 AD
CCNK	8812	AD	PMID: 30122539
CDC42	998	AD	PMID: 29394990
CDC42BPB	9578	AD	PMID:32031333
CDC6	990	AR	?Meier-Gorlin syndrome 5, 613805
CDH11	1009	AR	Elsahy-Waters syndrome, 211380
CDH15	1013	AD	Mental retardation, autosomal dominant 3, 612580
CDH2	1000	AD	PMID:31585109
CDK10	8558	AR	AR. AL KAISSI SYNDROME,617684
CDK13	8621	AD	Congenital heart defects dysmorphic facial features and intellectual development disorder 617360 OMIM:617360:Autosomal dominant Congenital heart defects dysmorphic facial features and intellectual development disorder 61736
CDK19	23097	AD	PMID:32330417
CDK5RAP2	55755	AR	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	1024	AD	PMID:30905399
CDKL5	6792	XLD	Epileptic encephalopathy, early infantile, 2, 300672
CDON	50937	AD	Holoprosencephaly 11, 614226

CENPJ	55835	AR	?Seckel syndrome 4, 613676|Microcephaly 6, primary, autosomal recessive, 608393
CEP104	9731	AR	Joubert syndrome 25, 616781
CEP120	153241	AR	Joubert syndrome 31, 617761
CEP135	9662	AR	?Microcephaly 8, primary, autosomal recessive, 614673
CEP152	22995	AR	Microcephaly 9, primary, autosomal recessive, 614852
CEP290	80184	AR	Joubert syndrome 5, 610188|Meckel syndrome 4, 611134
CEP41	95681	AR	Joubert syndrome 15, 614464
CEP57	9702	AR	Mosaic variegated aneuploidy syndrome 2, 614114
CEP83	51134	AR	Nephronophthisis 18, 615862
CERS1	10715	AR	OMIM616230
CERT1	10087	AD	Mental retardation, autosomal dominant 34, 616351 (COL4A3BP)
CHAMP1	283489	AD	Mental retardation, autosomal dominant 40, 616579
CHD1	1105	AD	Pilarowski-Bjornsson syndrome, 617682
CHD2	1106	AD	Epileptic encephalopathy, childhood-onset, 615369
CHD3	1107	AD	Snijders Blok-Campeau syndrome 618205
CHD4	1108	AD	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	55636	AD	CHARGE syndrome, 214800|Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	57680	AD	{Autism, susceptibility to, 18}, 615032
CHKB	1120	AR	Muscular dystrophy, congenital, megaconial type, 602541
CHL1	10752	AD	Palumbo et al Mol Cytogenet. 2015
CHMP1A	5119	AR	PONTOCEREBELLAR HYPOPLASIA AND MICROCEPHALY OMIM:614961
CIC	23152	AD	PMID:28288114, Mental retardation, autosomal dominant 45 617600 AD
CIT	11113	AR	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	150468	AR	Filippi syndrome, 272440
CLCN4	1183	XLR	Mental retardation, X-linked 49/15, 300114
CLIC2	1193	XLR	?Mental retardation, X-linked, syndromic 32, 300886
CLIP1	6249	AR	PMID:29302074
CLN3	1201	AR	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	1203	AR	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	54982	AR	Ceroid lipofuscinosis, neuronal, 6, 601780|Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	2055	AR	Ceroid lipofuscinosis, neuronal, 8, 600143|Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	10978	AR	Pontocerebellar hypoplasia, type 10, 615803
CLPB	81570	AR	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLTC	1213	AD	PMID 31776469
CNKSR2	22866	XL	Intellectual disability,X-linked non syndromic (Vaags (2014) Ann Neurol 76,758)
CNNM2	54805	AD/AR	Hypomagnesemia 6, renal, 613882
CNOT1	23019	AD	PMID:31006510; OMIM #618500
CNOT2	4848	AD	No OMIM phenotype
CNOT3	4849	AD	PMID 31201375
CNPY3	10695	AR	Epileptic encephalopathy, early infantile, 60, 617929
CNTNAP2	26047	AR	Cortical dysplasia-focal epilepsy syndrome, 610042|Pitt-Hopkins like syndrome 1, 610042
COA8	84334	AR	Mitochondrial complex IV deficiency, 220110
COASY	80347	AR	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COG1	9382	AR	Congenital disorder of glycosylation, type IIg, 611209
COG5	10466	AD	Congenital disorder of glycosylation, type IIi, 613612
COG6	57511	AR	Congenital disorder of glycosylation, type III, 614576|Shaheen syndrome, 615328

COG7	91949	AR	Congenital disorder of glycosylation, type IIe, 608779
COG8	84342	AR	Congenital disorder of glycosylation, type IIh, 611182
COL18A1	80781	AR	OMIM267750
COL4A1	1282	AD	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773|Brain small vessel disease with or without ocular anomalies, 607595|Porencephaly 1, 175780
COL4A2	1284	AD	Porencephaly 2, 614483
COLEC11	78989	AR	3MC syndrome 2, 265050
COPB1	1315	AR	PMID: 33632302
COQ2	27235	AR	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	51117	AR	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	56997	AR	Coenzyme Q10 deficiency primary 4 612016|Autosomal recessive Coenzyme Q1 deficiency primary 4 61216
COQ9	57017	AR	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	1352	AR	Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX15	1355	AR	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119|Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	1340	AR	Mitochondrial complex IV deficiency, 220110
CPLANE1	65250	AR	Joubert syndrome 17, 614615 (C5ORF42)
CPLX1	10815	AR	Epileptic encephalopathy, early infantile, 63, 617976
CPS1	1373	AR	Carbamoylphosphate synthetase I deficiency, 237300
CPT2	1376	AD/AR	24/03/2021 15:34
CRADD	8738	AR	Mental retardation, autosomal recessive 34, with variant lissencephaly 614499
CRBN	51185	AR	Mental retardation, autosomal recessive 2, 607417
CREBBP	1387	AD	Rubinstein-Taybi syndrome, 180849
CRPPA	729920	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (ISPD)
CSDE1	7812	AD	PMID:31579823
CSF1R	1436	AR	PMID:30982608; OMIM #618476
CSNK2A1	1457	AD	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	1460	AD	ID, poiret et alHum mut 2017
CSPP1	79848	AR	Joubert syndrome 21, 615636
CSTB	1476	AR	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	1487	AD	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome 617915 AD
CTC1	80169	AR	OMIM612199
CTCF	10664	AD	Mental retardation, autosomal dominant 21, 615502
CTDP1	9150	AR	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	1496	AR	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNB1	1499	AD	Colorectal cancer, somatic, 114500|Mental retardation, autosomal dominant 19, 615075
CTSA	5476	AR	Galactosialidosis, 256540
CTSD	1509	AR	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	83992	AD	?Autism (Iossifov (2012) Neuron 74,285)|Autism,Guo et al., Mol Psy 2018
CUL4B	8450	XLR	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX1	1523	AD	Global developmental delay with or without impaired intellectual development, 618330
CUX2	23316	AD	neurdevelopmental disorder,Geisheker et al nature NEUROSCIENCE 2017
CWC27	10283	AR	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	55280	AR	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXORF56	63932	XR	PMID: 29374277

CYB5R3	1727	AR	Methemoglobinemia, type I, 250800
CYFIP2	26999	AD	PMID:30664714; OMIM #618008
CYP27A1	1593	AR	Cerebrotendinous xanthomatosis, 213700
CYP2U1	113612	AR	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	9420	AR	Spastic paraplegia 5A autosomal recessive 278
D2HGDH	728294	AR	D-2-hydroxyglutaric aciduria, 600721
DAG1	1605	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS1	1615	AR	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	55157	AR	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	1629	AR	Maple syrup urine disease, type II, 248600
DCAF17	80067	AR	Woodhouse-Sakati syndrome, 241080
DCC	1630	AD/AR Mirror movements 1 and/or agenesis of the corpus callosum 157600	27/01/2020 8:48
DCHS1	8642	AR	Mitral valve prolapse 2, 607829
DCPS	28960	AR	Al-Raqad syndrome, 616459
DCX	1641	XL	Lissencephaly, X-linked, 300067
DDB1	1642	AD	33743206
DDC	1644	AR	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD2	23259	AR	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	1650	AR	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IR; CDG1R, OMIM:614507(Global developmental delay)
DDX11	1663	AR	Warsaw breakage syndrome, 613398
DDX3X	1654	XLD	Mental retardation, X-linked 102, 300958
DDX59	83479	AR	Orofaciodigital syndrome V, 174300
DDX6	1656	AD	PMID:31422817; OMIM #618653
DEAF1	10522	AD/AR	Mental retardation, autosomal dominant 24, 615828
DEGS1	8560	AR	PMID:31186544; OMIM #618404
DENND5A	23258	AR	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	9681	AD	OMIM604364
DHCR24	1718	AR	Desmosterolosis, 602398
DHCR7	1717	AR	Smith-Lemli-Opitz syndrome, 270400
DHDDS	79947	AD	Developmental delay and seizures with or without movement abnormalities 617836 AD
DHFR	1719	AR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	1725	AR	PMID:30661771; OMIM #618480
DHTKD1	55526	AR	2-aminoadipic 2-oxoadipic aciduria, 204750|?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	22907	AD	microcephaly, developmental delay intellectual disability, mild cerebral volume loss, hypotonia, seizures, short stature, failure to thrive, and generalized hirsutism (GenomicsEngland,
DIP2B	57609	AD	Mental retardation, FRA12A type, 136630
DIS3L2	129563	AR	Perlman syndrome, 267000
DKC1	1736	XLR	Dyskeratosis congenita, X-linked, 305000
DLD	1738	AR	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	1741	XL	Mental retardation, X-linked 90, 300850
DLG4	1742	AD	PMID: 29460436
DLL1	28514	AD	PMID:31353024
DMD	1756	XLR	Becker muscular dystrophy, 300376|Duchenne muscular dystrophy, 310200
DMPK	1760	AD	Myotonic dystrophy 1, 160900
DMXL2	23312	AR	OMIM618663

DNAJA1	3301	AR	PMID:30972502
DNAJC12	56521	AR	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	131118	AR	3-methylglutaconic aciduria, type V, 610198
DNM1	1759	AD	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	10059	AD/AR	OMIM614388
DNMT3A	1788	AD	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	1789	AR	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK3	1795	AR	OMIM618292
DOCK6	57572	AR	Adams-Oliver syndrome 2, 614219
DOCK7	85440	AR	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	81704	AD	AD.Mental retardation autosomal dominant 2 614113
DOLK	22845	AR	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	1798	AD/AR	Congenital disorder of glycosylation, type Ij, 608093|Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	5977	AD	Coffin-Siris syndrome 7 618027 AD
DPH1	1801	AR	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	8813	AR	Congenital disorder of glycosylation, type Ie, 608799
DPP6	1804	AD	Mental retardation, autosomal dominant 33, 616311
DPYD	1806	AR	5-fluorouracil toxicity, 274270|Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	1807	AR	Dihydropyrimidinuria, 222748
DYM	54808	AR	Dyggve-Melchior-Clausen disease, 223800|Smith-McCort dysplasia, 607326
DYNC1H1	1778	AD	Charcot-Marie-Tooth disease, axonal, type 20, 614228|Mental retardation, autosomal dominant 13, 614563
DYNC1I2	1781	AR	PMID:31079899; OMIM #618492
DYRK1A	1859	AD	Mental retardation, autosomal dominant 7, 614104
EBF3	253738	AD	Intellectual Disability, Ataxia, and Facial Dysmorphism, OMIM:617330(Cerebellar atrophy|Delayed speech and language development|Global developmental delay|Intellectual disability)
EBP	10682	XLR/XLD	Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	1892	AR	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EED	8726	AD	Cohen-Gibson syndrome, 617561
EEF1A2	1917	AD	Epileptic encephalopathy, early infantile, 33, 616409|Mental retardation, autosomal dominant 38, 616393
EEF1B2	1933	AR	AUTOSOMAL RECESSIVE MENTAL RETARDATION (GenomicsEngland,
EFTUD2	9343	AD	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	79813	AD	Kleefstra syndrome, 610253
EIF2A	83939	AR	PMID:31130284
EIF2AK1	27102	AD	PMID:32197074
EIF2AK2	5610	AD	PMID:32197074
EIF2AK3	9451	AR	Wolcott-Rallison syndrome, 226980
EIF2S3	1968	XLR	Mental retardation X-linked syndromic Borck type 3987
EIF3F	8665	AR	Mental retardation, autosomal recessive 67, 618295
EIF4A3	9775	AR	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	60528	AR	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELOVL4	6785	AD	Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP2	55250	AR	Mental retardation, autosomal recessive 58, 617270
EMC1	23065	AR	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EML1	2009	AR	Band heterotopia, 600348
EMX2	2018	AD	Schizencephaly, 269160
ENTPD1	953	AR	Spastic paraplegia 64, autosomal recessive, 615683
EP300	2033	AD	Colorectal cancer, somatic, 114500|Rubinstein-Taybi syndrome

			2, 613684
EPG5	57724	AR	Vici syndrome, 242840
EPM2A	7957	AR	OMIM254780
EPRS1	2058	AR	PMID: 29576217
ERCC1	2067	AR	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	2068	AR	Cerebrooculofacioskeletal syndrome 2, 610756|Trichothiodystrophy 1, photosensitive, 601675
ERCC3	2071	AR	Trichothiodystrophy 2, photosensitive, 616390
ERCC5	2073	AR	Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	2074	AR	Cerebrooculofacioskeletal syndrome 1, 214150|Cockayne syndrome, type B, 133540|De Sanctis-Cacchione syndrome, 278800
ERCC8	1161	AR	Cockayne syndrome, type A, 216400
ERLIN2	11160	AR	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	157570	AR	Roberts syndrome, 268300|SC phocomelia syndrome, 269000
ETFB	2109	AR	Glutaric acidemia IIB, 231680
ETHE1	23474	AR	Ethylmalonic encephalopathy, 602473
EXOSC2	23404	AR	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	51010	AR	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	56915	AR	PMID:32504085
EXOSC9	5393	AR	PMID: 29727687
EXTL3	2137	AR	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	2146	AD	Weaver syndrome, 277590
FA2H	79152	AR	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	84668	AR	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	317662	AR	PMID: 30905400
FAM20C	56975	AR	Raine syndrome, 259775
FAM50A	9130	XLR	PMID:32703943
FANCL	55120	AR	Fanconi anemia, complementation group L, 614083
FAR1	84188	AR	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	10667	AR	OMIM614946
FAT4	79633	AR	Van Maldergem syndrome 2, 615546
FBN1	2200	AD	AD.MARFAN SYNDROME (MFS), 614185
FBRSL1	57666	AD	PMID:32424618
FBXL3	26224	AR	PMID: 30481285
FBXL4	26235	AR	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	80204	AD	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities 618089
FBXO31	79791	AR	?Mental retardation, autosomal recessive 45, 615979
FBXW11	23291	AD	PMID:31402090
FCSK	197258	AR	PMID: 30503518 (FUK)
FGD1	2245	XLR	Aarskog-Scott syndrome, 305400|Mental retardation, X-linked syndromic 16, 305400
FGF12	2257	AD	Epileptic encephalopathy, early infantile, 47 617166 AD
FGF13	2258	XL(D?)	PMID:33245860 (alt name FHF2)
FGF14	2259	AD	Spinocerebellar ataxia 27, 609307
FGFR1	2260	AD	Encephalocraniocutaneous lipomatosis,613001|Hartsfield syndrome, 615465|Trigonocephaly 1, 190440
FGFR2	2263	AD	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410
FGFR3	2261	AR	Achondroplasia, 100800
FH	2271	AR	Fumarase deficiency, 606812|Leiomyomatosis and renal cell cancer, 150800
FIBP	9158	AR	Thauvin-Robinet-Faivre syndrome, 617107
FKRP	79147	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain

			and eye anomalies), type A, 5, 613153|Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5,606612)
FKTN	2218	AR	Cardiomyopathy, dilated, 1X, 611615|Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152
FLNA	2316	XL	Cardiac valvular dysplasia, X-linked, 314400|Frontometaphyseal dysplasia, 305620
FLVCR1	28982	AR	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	55640	AR	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	56776	AR	Mental retardation, autosomal recessive 47, 616193
FMR1	2332	XLD	Fragile X syndrome, 300624
FOLR1	2348	AR	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	2290	AD	Rett syndrome, congenital variant, 613454
FOXP1	27086	AD	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	93986	AD	Speech-language disorder-1, 602081
FOXP4	116113	AD	PMID:33110267
FOXRED1	55572	AR	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	80144	AR	Fraser syndrome, 219000
FRMPD4	9758	XLR	Mental retardation,X-linked 104, 300983
FRRS1L	23732	AR	Epileptic encephalopathy, early infantile, 37, 616981
FTCD	10841	AR	Glutamate formiminotransferase deficiency, 229100
FTO	79068	AR	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	24140	XLR	Mental retardation, X-linked 9, 309549
FUCA1	2517	AR	Fucosidosis, 230000
FUT8	2530	AR	Congenital disorder of glycosylation with defective fucosylation 1, 618005
GABBR2	9568	AD	EPILEPTIC ENCEPHALOPATHY
GABRA1	2554	AD	Epileptic encephalopathy, early infantile, 19, 615744
GABRA2	2555	AD	OMIM618557
GABRA3	2556	XL	PMID: 29053855
GABRB1	2560	AD	OMIM617153
GABRB2	2561	AD	PMID: 29942082
GABRB3	2562	AD	{Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	2566	AD	Febrile seizures, familial, 8, # 611277
GAD1	2571	AR	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	2581	AR	Krabbe disease, 245200
GALE	2582	AR	Galactose epimerase deficiency, 230350
GALNT2	2590	AR	PMID:32293671
GALT	2592	AR	Galactosemia, 230400
GAMT	2593	AR	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	57459	AD	Mental retardation, autosomal dominant 18, 615074
GATM	2628	AR	Cerebral creatine deficiency syndrome 3, 612718
GBA	2629	AR	GAUCHER DISEASE
GCDH	2639	AR	Glutaricaciduria, type I, 231670
GCH1	2643	AR	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCSH	2653	AR	Glycine encephalopathy, 605899
GDI1	2664	XL	Mental retardation, X-linked 41, 300849
GFAP	2670	AD	Alexander disease, 203450
GFM1	85476	AR	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	84340	AR	Combined oxidative phosphorylation deficiency 39, 618397
GJA1	2697	AD	PMID:19808103
GJB1	2705	XLR	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1,

			OMIM:302800(Abnormality of the cerebral white matter|Motor delay|Cerebellar atrophy)
GJC2	57165	AR	Leukodystrophy, hypomyelinating, 2, 608804|Spastic paraplegia 44, autosomal recessive, 613206
GK	2710	XLD	Glycerol kinase deficiency, 307030
GLB1	2720	AR	GM1-gangliosidosis, type I, 230500
GLDC	2731	AR	Glycine encephalopathy, 605899
GLI2	2736	AD	Culler-Jones syndrome, 615849|Holoprosencephaly-9, 610829
GLI3	2737	AD	Greig cephalopolysyndactyly syndrome, 175700|Pallister-Hall syndrome, 146510
GLIS3	169792	AR	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLUD1	2746	AD	# 606762. HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 6; HHF6
GLYCTK	132158	AR	D-glyceric aciduria, 220120
GM2A	2760	AR	GM2-gangliosidosis, AB variant, 272750
GMPPA	29926	AR	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	29925	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350|Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351
GNAI1	2770	AD	GNAI1 syndrome (GenomicsEngland,
GNAO1	2775	AD	Epileptic encephalopathy, early infantile, 17, 615473
GNAS	2778	AD	Acromegaly, somatic, 102200
GNB1	2782	AD	Leukemia,acute lymphoblastic,somatic, 613065|Mental retardation, autosomal dominant 42, 616973
GNB5	10681	AR	Intellectual developmental disorder with cardiac arrhythmia, 617173|Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNPAT	8443	AR	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	79158	AR	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	84572	AR	Mucolipidosis III gamma, 252605
GNS	2799	AR	Mucopolysaccharidosis type IIID, 252940
GOSR2	9570	AR	OMIM614018
GOT2	2806	AR	PMID:31422819
GPAA1	8733	AR	Glycosylphosphatidylinositol biosynthesis defect 15 617810(global developmental delay)
GPC3	2719	XLR	Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	2239	XR	PMID:30982611
GPHN	10243	AR	Molybdenum cofactor deficiency C, 615501
GPSM2	29899	AR	Chudley-McCullough syndrome, 604213
GPT2	84706	AR	?Mental retardation, autosomal recessive 49, 616281
GRIA1	2890	AD	neurdevelopmental disorder,Geisheker et al nature NEUROSCIENCE 2017
GRIA2	2891	AD	Autism, Guo et al., Mol Psy 2018
GRIA3	2892	XLR	Mental retardation, X-linked 94, 300699
GRIA4	2893	AD	Neurodevelopmental disorder with or without seizures and gait abnormalities.#617864
GRID2	2895	AR	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	2898	AR	Mental retardation, autosomal recessive, 6, 611092
GRIN1	2902	AD	Mental retardation, autosomal dominant 8, 614254
GRIN2A	2903	AD	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	2904	AD	Epileptic encephalopathy, early infantile, 27, 616139|Mental retardation, autosomal dominant 6, 613970
GRIN2D	2906	AD	Epileptic encephalopathy, early infantile, 46, OMIM:617162(Epileptic encephalopathy|Global developmental delay|Hypsarrhythmia)

GRIPAP1	56850	AD	PMID: 28285821 (GRASP1)
GRM1	2911	AR	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSS	2937	AR	Glutathione synthetase deficiency, 266130|Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	2961	AR	Trichothiodystrophy 6, nonphotosensitive, OMIM:616943(Intellectual disability, moderate|Motor delay)
GTF2H5	404672	AR	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	54676	AR	JABERI-ELAHI SYNDROME,OMIM 617988
GTPBP3	84705	AR	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	2990	AR	Mucopolysaccharidosis VII, 253220
H1-4	3008	AD	Rahman syndrome 617537; mild to severe intellectual disability OMIM:617537 (HIST1H1E)
H4C3	8364	AD	PMID:28920961 (alt nameHIST1H4C)
HACE1	57531	AR	PMID:26437029
HADH	3033	AR	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	3030	AR	Fatty liver, acute, of pregnancy, 609016
HAX1	10456	AR	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	3052	XLD	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	3054	XL	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	348980	AD	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	9759	AD	Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706)|Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219)|?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HDAC6	10013	XLD	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia,300863
HDAC8	55869	XL	Cornelia de Lange syndrome 5, 300882
HECW2	57520	AD	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	220296	AR/AD	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925
HERC1	8925	AR	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	8924	AR	Mental retardation, autosomal recessive 38, 615516
HESX1	8820	AR	Growth hormone deficiency with pituitary anomalies, 182230
HEXA	3073	AR	GM2-gangliosidosis, several forms, 272800|Tay-Sachs disease, 272800
HEXB	3074	AR	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	138050	AR	MUCOPOLYSACCHARIDOSIS TYPE 3C (MPS3C), OMIM:252930(Intellectual disability|Motor delay)
HIBCH	26275	AR	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIVEP2	3097	AD	Mental retardation, autosomal dominant 43, 616977
HK1	3098	AD	PMID:30778173; OMIM #618547
HLCS	3141	AR	Holocarboxylase synthetase deficiency, 253270
HMGCL	3155	AR	HMG-CoA lyase deficiency, 246450
HNMT	3176	AR	PMID:29302074
HNRNPH1	3187	AD	PMID: 29938792
HNRNPH2	3188	XLD	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	3190	AD	Au-Kline syndrome, 616580
HNRNPR	10236	AD	PMID:31079900
HNRNPU	3192	AD	Epileptic encephalopathy, early infantile, 54, 617391
HOXA1	3198	AR	Athabaskan brainstem dysgenesis syndrome, 601536|Bosley-Salih-Alorainy syndrome, 601536
HPD	3242	AR	Hawkinsinuria, 140350
HPDL	84842	AR	PMID:32707086
HPRT1	3251	XLR	HPRT-related gout, 300323|Lesch-Nyhan syndrome, 300322
HRAS	3265	AD	Congenital myopathy with excess of muscle spindles, 218040|Costello syndrome, 218040|Schimmelpenning-

			Feuerstein-Mims syndrome, somatic mosaic, 163200
HS2ST1	9653	AR	PMID:33159882
HSD17B10	3028	XL	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438|?Mental retardation, X-linked syndromic 10, 300220
HSD17B4	3295	AR	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPA9	3313	AD	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	3329	AD/AR	Leukodystrophy, hypomyelinating, 4, 612233|Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	27429	AR	3-methylglutaconic aciduria, type VIII, 617248
HUWE1	10075	XL	Mental retardation, X-linked syndromic, Turner type, 300706
HYLS1	219844	AR	Hydrolethalus syndrome, 236680
IARS1	3376	AR	Growth retardation, intellectual developmental disorder, hypotonia and hepatopathy, 617093
ICE1	23379	AR	PMID:31130284
IDS	3423	XLR	Mucopolysaccharidosis II, 309900
IDUA	3425	AR	Mucopolysaccharidosis Ih, 607014
IER3IP1	51124	AR	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	64135	AD	# 615846. AICARDI-GOUTIERES SYNDROME 7; AGS7
IFT172	26160	AR	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IGBP1	3476	XLR	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	3479	AR	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	3480	AR	Insulin-like growth factor I, resistance to, 270450
IGSF1	3547	XLR	Hypothyroidism central and testicular enlargement, 300888
IKBKG	8517	XLD	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291
IL1RAPL1	11141	XLR	Mental retardation, X-linked 21/34, 300143
INPP5E	56623	AR	Joubert syndrome 1, 213300|Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	51763	AR	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INTS1	26173	AR	PMID:30622326; OMIM #618571
IQSEC1	9922	AR	PMID:31607425
IQSEC2	23096	XLD	Mental retardation, X-linked 1/78, 309530
IQSEC3	440073	AR	PMID: 31130284
IRF2BPL	64207	AD	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures 618088
ITGA7	3679	AR	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	3704	AR	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	3708	AR	Gillespie syndrome,206700|Spinocerebellar ataxia 15, 606658|Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	3712	AR	Isovaleric acidemia, 243500
JAG1	182	AD	Alagille syndrome, 118450|Tetralogy of Fallot, 187500
JAM3	83700	AR	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730|?Rett syndrome (Saez (2016) Genet Med 18,378)|?Congenital heart disease in 22q11.2 deletion syndrome patients (Guo (2015) Am J Hum Genet 97,869)|?Autism spectrum disorder (Saez (2016) Genet Med 18,378)
JARID2	3720	AD	PMID:33077894
JMJD1C	221037		No OMIM phenotype
KANK1	23189	AD with maternal imprinting	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	284058	AD	Koolen-De Vries syndrome, 610443
KARS1	3735	AR	OMIM619147

KAT5	10524	AD	PMID:32822602
KAT6A	7994	AD	Mental retardation, autosomal dominant 32, 616268
KAT6B	23522	AD	Genitopatellar syndrome, 606170
KATNB1	10300	AR	Lissencephaly 6, with microcephaly, 616212
KCNA1	3736	AD	OMIM160120
KCNA2	3737	AD	# 616366. EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 32; EIEE32
KCNB1	3745	AD	# 616056. EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 26; EIEE26
KCNC1	3746	AD	EPILEPSY PROGRESSIVE MYOCLONIC 7 OMIM:616187
KCNC3	3748	AD	Spinocerebellar ataxia 13, 605259
KCND3	3752	AD	Spinocerebellar ataxia 19, OMIM:607346(Cerebellar atrophy)
KCNH1	3756	AD	Temple-Baraitser syndrome, 611816|Zimmermann-Laband syndrome 1, 135500
KCNJ10	3766	AR	Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	3767	AD	Diabetes mellitus, permanent neonatal, with neurologic features, 606176
KCNJ6	3763	AD	Keppen-Lubinsky syndrome, 614098
KCNK4	50801	AD	PMID: 30290154
KCNK9	51305	AD	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	3778	AD	PMID: 31152168
KCNN3	3782	AD	PMID: 31155282; OMIM #618658
KCNQ2	3785	AD	Epileptic encephalopathy, early infantile, 7, 613720
KCNQ3	3786	AD	Seizures, benign neonatal, type 2, 121201
KCNQ5	56479	AD	Mental retardation, autosomal dominant 46 617601
KCNT1	57582	AD	Epilepsy, nocturnal frontal lobe, 5, 615005|Epileptic encephalopathy, early infantile, 14, 614959
KCNT2	343450	AD	OMIM617771
KCTD7	154881	AR	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	23028	AD	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	51780	AD	PMID: 30929739
KDM4B	23030	AD	PMID:33232677
KDM5B	10765	AD	neurodevelopment delay and autism spectrum disorder (GenomicsEngland,
KDM5C	8242	XLR	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	7403	XLD	Kabuki syndrome 2, 300867
KDM6B	23135	AD	Neurodevelopmental delay and dysmorphic features
KIAA0586	9786	AR	Joubert syndrome 23, 616490
KIAA1109	84162	ad	Dandy-Walker malformation,hydrocephalus,flexed deformity,club feet,micrognathia and pleural effusion (Alazami (2015) Cell Rep 10,148)
KIDINS220	57498	AD	# 617296. SPASTIC PARAPLEGIA, INTELLECTUAL DISABILITY, NYSTAGMUS, AND OBESITY; SINO
KIF11	3832	AD	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	9928	AR	?Meckel syndrome 12, 616258
KIF1A	547	AD/AR	Mental retardation, autosomal dominant 9, 614255|Spastic paraplegia 30, autosomal recessive, 610357
KIF2A	3796	AD	OMIM615411
KIF4A	24137	XLR	?Mental retardation, X-linked 100, 300923
KIF5C	3800	AD	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	374654	AD/AR	Acrocallosal syndrome, 200990|Joubert syndrome 12, 200990
KIFBP	26128	AR	Goldberg-Shprintzen megacolon syndrome OMIM:609460
KIRREL3	84623	AD	Mental retardation, autosomal dominant 4, 612581
KLF7	8609	AD	PMID: 29251763

KLHL15	80311	XLR	Mental retardation,X-linked 103,300982
KLHL7	55975	AD/AR	COLD-INDUCED SWEATING SYNDROME 3 (AR), 617055| RETINITIS PIGMENTOSA 42,612943:(AD)
KMT2A	4297	AD	Leukemia, myeloid/lymphoid or mixed-lineage, 159555|Wiedemann-Steiner syndrome, 605130
KMT2B	9757	AD	DYSTONIA 28, CHILDHOOD-ONSET; 617284
KMT2C	58508	AD	Kleefstra syndrome (Kleefstra (2012) Am J Hum Genet 91,73)
KMT2D	8085	AD	Kabuki syndrome 1, 147920
KMT2E	55904	AD	O
KMT5B	51111	AD	Mental retardation autosomal dominant 51.# 617788
KNL1	57082	AR	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	11133	AR	Mental retardation, autosomal recessive 41, 615637
KRAS	3845	AD	Bladder cancer, somatic, 109800
L1CAM	3897	XLR	Corpus callosum, partial agenesis of, 304100|CRASH syndrome, 303350
L2HGDH	79944	AR	L-2-hydroxyglutaric aciduria, 236792
LAMA1	284217	AR	Poretti-Boltshauser syndrome, 615960
LAMA2	3908	AR	Muscular dystrophy, congenital merosin-deficient, 607855
LAMB1	3912	AR	Lissencephaly 5, 615191
LAMC3	10319	AR	Cortical malformations, occipital, 614115
LAMP2	3920	XLD	Danon disease, 300257
LARGE1	9215	AR	AR.Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6 613154|Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 6 6884
LARP7	51574	AR	Alazami syndrome, 615071
LAS1L	81887	XLR	Wilson-Turner syndrome, 309585
LGI4	163175	AR	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LIAS	11019	AR	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	3981	AR	LIG4 syndrome, 606593
LINGO1	84894	AR	PMID: 28837161
LINS1	55180	AR	AUTOSOMAL RECESSIVE MENTAL RETARDATION, OMIM:614340(Global developmental delay|Poor speech|Intellectual disability)
LMAN2L	81562	AR	?Mental retardation, autosomal recessive, 52, 616887
LMNB1	4001	AD	PMID:32910914
LONP1	9361	AR	CODAS syndrome, 600373
LRP2	4036	AR	Donnai-Barrow syndrome, 222448
LRPPRC	10128	AR	Leigh syndrome, French-Canadian type, 220111
LSS	4047	AR	PMID:30723320
LZTFL1	54585	AR	Bardet-Biedl syndrome 17, 615994
LZTR1	8216	AD/AR	# 616564. NOONAN SYNDROME 10 / #605275 Noonan syndrome 2
MAB21L1	4081	AR	PMID: 30487245
MAB21L2	10586	AD	PMID: 26116559
MACF1	23499	AD	PMID: 30471716
MADD	8567	AR	PMID:32761064
MAF	4094	AD	Ayme-Gripp syndrome 601088
MAG	4099	AR	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	54551	AD	Schaaf-Yang syndrome, 615547
MAGT1	84061	XL	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAN1B1	11253	AR	Mental retardation, autosomal recessive 15, 614202
MAN2B1	4125	AR	Mannosidosis, alpha-, types I and II, 248500
MANBA	4126	AR	Mannosidosis, beta, 248510
MAOA	4128	XLR	Brunner syndrome, 300615
MAP1B	4131	AD	PMID: 30150678

MAP2K1	5604	AD	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	5605	AD	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	5594	AD	PMID:32721402
MAPK8IP3	23162	AD	PMID:30612693; OMIM #618443
MAPRE2	10982	AR	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	5648	AR	3MC syndrome 1, 257920
MAST1	22983	AD	PMID: 31721002
MAT1A	4143	AD	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MBD5	55777	AD	Mental retardation, autosomal dominant 1, 156200
MBOAT7	79143	AR	Mental retardation, autosomal recessive 57, 617188
MBTPS2	51360	XLR	IFAP syndrome with or without BRESHECK syndrome, 308205
MCCC1	56922	AR	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	64087	AR	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	57192	AR	Mucolipidosis IV, 252650
MCPH1	79648	AR	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	4191	AR	Epileptic encephalopathy, early infantile, 51, 617339
MECP2	4204	XL	Encephalopathy, neonatal severe, 300673|Mental retardation, X-linked syndromic, Lubs type, 300260|Mental retardation, X-linked, syndromic 13, 300055|Rett syndrome, 312750
MED12	9968	XLR	Lujan-Fryns syndrome, 309520|Ohdo syndrome, X-linked, 300895|Opitz-Kaveggia syndrome, 305450
MED13	9969	AD	Snijders Blok 2018
MED13L	23389	AD	Mental retardation and distinctive facial features with or without cardiac defects, 616789
MED17	9440	AR	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	9439	AR	Mental retardation, autosomal recessive 18, 614249
MED25	81857	AR	?Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	4208	AD	Chromosome 5q14.3 deletion syndrome, 613443|Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	1954	AR	Carpenter syndrome 2, 614976
MEIS2	4212	AD	Heart defect, cleft palate and intellectual disability (Louw (2015) Am J Med Genet A 167, 1142)|Orofacial clefting & delayed motor development (Johansson (2014) Am J Med Genet A 164, 1622)
METTL23	124512	AR	Mental retardation, autosomal recessive 44, 615942
METTL5	29081	AR	PMID:31564433; OMIM #618665
MFF	56947	AR	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	84879	AR	PMID:29302074
MFSD8	256471	AR	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	4247	AR	Congenital disorder of glycosylation, type IIa, 212066
MGP	4256	AR	Keutel syndrome, 245150
MICU1	10367	AR	Myopathy with extrapyramidal signs, 615673
MID1	4281	XLR	Opitz GBBB syndrome, type I, 300000
MKKS	8195	AR	Bardet-Biedl syndrome 6, 605231|McKusick-Kaufman syndrome, 236700
MKS1	54903	AR	Bardet-Biedl syndrome 13, 615990
MLC1	23209	AR	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLYCD	23417	AR	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	166785	AR	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	326625	AR	METHYLMALONIC ACIDURIA TYPE CBLB, OMIM:251110(Global developmental delay)
MMACHC	25974	AR	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	27249	AR	Homocystinuria, cblD type, variant 1, 277410
MMUT	4594	AR	Methylmalonic aciduria, (mut) type, 251000
MN1	4330	AD	Autism Spectrum Disorders,Pubmed

			31834374|OMIM:607174:Meningioma
MOCS1	4337	AR	Molybdenum cofactor deficiency A, 252150
MOCS2	4338	AR	Molybdenum cofactor deficiency B, 252160
MOGS	7841	AR	Congenital disorder of glycosylation, type IIb, 606056
MORC2	22880	AD	PMID:32693025
MOV10	4343	AR	PMID:31130284
MPDU1	9526	AR	Congenital disorder of glycosylation, type If, 609180
MPDZ	8777	AR	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPLKIP	136647	AR	Trichothiodystrophy 4, nonphotosensitive, 234050
MRPS22	56945	AR	Combined oxidative phosphorylation deficiency 5, 611719
MSL3	10943	XL	DDD-study + PMID 30224647
MSMO1	6307	AR	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTFMT	123263	AR	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	4524	AR	Homocystinuria due to MTHFR deficiency, 236250
MTOR	2475	AD	Smith-Kingsmore syndrome 616638 AD
MTR	4548	AR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	4552	AR	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MVK	4598	AR	Hyper-IgD syndrome, 260920
MYCN	4613	AD	Feingold syndrome, 164280
MYH9	4627	AD	Deafness, autosomal dominant 17, 603622|Epstein syndrome, 153650|Fechtner syndrome, 153640|Macrothrombocytopenia and progressive sensorineural deafness, 600208
MYO5A	4644	AR	Griscelli syndrome, type 1, 214450
MYT1L	23040	AD	Mental retardation, autosomal dominant 39, 616521
NAA10	8260	XLD	Ogden syndrome, 300855
NAA15	80155	AD	Mental retardation, autosomal dominant 50 617787
NACC1	112939	AD	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties and delayed brain myelination, 617393
NAGA	4668	AR	Kanzaki disease, 609242|Schindler disease, type I, 609241
NAGLU	4669	AR	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920|?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NALCN	259232	AD/AR	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANS	54187	AR	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS1	4677	AD/AR	PMID:32738225
NARS2	79731	AR	Combined oxidative phosphorylation deficiency 24, 616239
NAXD	55739	AR	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2 618321
NBEA	26960	AD	PMID: 30269351
NBN	4683	AR	Aplastic anemia, 609135|Nijmegen breakage syndrome, 251260
NCAPD2	9918	AR	PMID:31056748
NCAPG2	54892	AR	PMID:30609410; OMIM #618460
NCKAP1	10787	AD	PMID:33157009
NDE1	54820	AR	Lissencephaly 4 (with microcephaly), 614019
NDP	4693	XLR	Exudative vitreoretinopathy 2, X-linked, 305390|Norrie disease, 310600
NDST1	3340	AR	Mental retardation, autosomal recessive 46, 616116
NDUFA1	4694	XLD	Mitochondrial complex I deficiency, 252010
NDUFA11	126328	XLD	Mitochondrial complex I deficiency, 252010
NDUFA12	55967	Mitochondrial	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	4695	AR	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	4700	AR	PMID: 30245030
NDUFAF3	25915	AR	Mitochondrial complex I deficiency, nuclear type 18, 618240

NDUFAF5	79133	AR	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	284184	AR	PMID:31866046
NDUFS1	4719	AR	Mitochondrial complex I deficiency, 252010
NDUFS2	4720	XLD	Mitochondrial complex I deficiency, 252010
NDUFS3	4722	AR	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFS4	4724	AR	Leigh syndrome, 256000
NDUFS7	374291	AR	Leigh syndrome, 256000
NDUFS8	4728	AR	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	4723	AR	Mitochondrial complex I deficiency, 252010
NDUFV2	4729	AR	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	23327	AD	Periventricular nodular heterotopia 7, 617201
NEU1	4758	AR	Sialidosis, type I, 256550
NEXMIF	340533	XLD	Mental retardation, X-linked 98, 300912
NF1	4763	AD	Leukemia, juvenile myelomonocytic, 607785|Neurofibromatosis, type 1, 162200
NF2	4771	AD	OMIM101000
NFIA	4774	AD	Brain malformation and urinary tract defect (Negishi (2015) Hum Genome Var 2)|Bipolar disorder and depression (Mikhail (2011) Am J Med Genet A 155,2386)|Central nervous system malformations (Koehler (2010) Eur J Pediatr 169,463)
NFIB	4781	AD	Macrocephaly, acquired, with impaired intellectual development 618286
NFIX	4784	AD	Marshall-Smith syndrome, 602535|Sotos syndrome 2, 614753
NFU1	27247	AR	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	55768	AR	Congenital disorder of deglycosylation, 615273
NHLRC1	378884	AR	OMIM254780
NHS	4810	XLD	Cataract 40, X-linked, 302200|Nance-Horan syndrome, 302350
NIPA1	123606	AD	OMIM600363
NIPBL	25836	AD	Cornelia de Lange syndrome 1, 122470
NKAP	79576	XR	PMID:31587868
NKX2-1	7080	AD	Choreoathetosis, hypothyroidism, and neonatal respiratory distress 610978 AD 3
NLGN3	54413	XL	{Asperger syndrome susceptibility, X-linked 1}, 300494
NLGN4X	57502	XLD	Mental retardation, X-linked, 300495
NONO	4841	XLR	Mental retardation, X-linked, syndromic 34, 300967
NOVA2	4858	AD	PMID:32197073
NPC1	4864	AR	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	10577	AR	Niemann-pick disease, type C2, 607625
NPHP1	4867	AR	Joubert syndrome 4, 609583
NPRL2	10641	AD	OMIM617116
NPRL3	8131	AD	OMIM617118
NR1I3	9970	AD	kleefstra et al, 2012 EHMT1-LIKE INTELLECTUAL DISABILITY
NR2F1	7025	AD	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	4929	AD	PMID: 31428396
NRAS	4893	AD	Colorectal cancer, somatic, 114500|Noonan syndrome 6, 613224
NRROS	375387	AR	PMID:32197075
NRXN1	9378	AR	Pitt-Hopkins-like syndrome 2, 614325
NSD1	64324	AD	Beckwith-Wiedemann syndrome, 130650|Sotos syndrome 1, 117550
NSD2	7468	AD	Wolf-Hirschhorn syndrome, OMIM:194190(Global developmental delay|Intellectual disability, severe|Agenesis of corpus callosum)
NSDHL	50814	XL	CHILD syndrome, 308050
NSUN2	54888	AR	Mental retardation, autosomal recessive 5, 611091
NT5C2	22978	AR	Spastic paraplegia 45, autosomal recessive, 613162

NTNG2	84628	AR	PMID:31668703
NTRK1	4914	AR	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	4915	AD	OMIM 617830 and 613886
NUBPL	80224	AR	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP62	23636	AR	Striatonigral degeneration, infantile, 271930
NUS1	116150	AD	Mental retardation, autosomal dominant 55, with seizures,617831
OCLN	1.01E+08	AR	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	4952	XLR	Dent disease 2, 300555|Lowe syndrome, 309000
ODC1	4953	AD	PMID: 30475435
OFD1	8481	XL	Joubert syndrome 10, 300804
OGT	8473	XL	Mental retardation X-linked 16 3997
OPHN1	4983	XLR	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	4998	AR	Meier-Gorlin syndrome 1, 224690
OSGEP	55644	AR	Galloway-Mowat syndrome 3, 617729
OTC	5009	XLR	Ornithine transcarbamylase deficiency, 311250
OTUD5	55593	XLR	PMID:33131077
OTUD6B	51633	AR	Intellectual developmental disorder with dysmorphic facies seizures and distal limb anomalies 617452
OTX2	5015	AD	OMIM610125
OXR1	55074	XMR	PMID:31785787
P4HTM	54681	AR	PMID: 30940925
PACS1	55690	AD	Schuss-Hoeijmakers-syndrome, 615009
PACS2	23241	AD	Epileptic encephalopathy, early infantile, 66 618067
PAFAH1B1	5048	AD	Lissencephaly 1, 607432|Subcortical laminar heterotopia, 607432
PAH	5053	AR	Phenylketonuria, 261600|[Hyperphenylalaninemia, non-PKU mild], 261600
PAK1	5058	AD	PMID: 30290153
PAK3	5063	XLR	Mental retardation, X-linked 30/47, 300558
PANK2	80025	AR	HARP syndrome, 607236|Neurodegeneration with brain iron accumulation 1, 234200
PARN	5073	AR	Dyskeratosis congenita, autosomal recessive 6, 616353
PARS2	25973	AR	OMIM618437
PAX1	5075	AR	?Otofaciocervical syndrome 2, 615560
PAX6	5080	AD/AR	Aniridia, 106210
PAX8	7849	AD	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	5087	AD	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay 617641 AD
PC	5091	AR	Pyruvate carboxylase deficiency, 266150
PCCA	5095	AR	Propionicacidemia, 606054
PCCB	5096	AR	Propionicacidemia, 606054
PCDH12	51294	AR	OMIM251280
PCDH19	57526	XL	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	7703	AD	INTELLECTUAL DUSBILITY (GenomicsEngland,
PCNT	5116	AR	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PDE4D	5144	AD	Acrodysostosis 2, with or without hormone resistance, 614613|{Stroke, susceptibility to, 1}, 606799
PDHA1	5160	XLD	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	8050	AR	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	54704	AR	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	23590	AR	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	57107	AR	Coenzyme Q10 deficiency, primary, 3, 614652

PEPD	5184	AR	Prolidase deficiency, 170100
PET100	1E+08	AR	Mitochondrial complex IV deficiency, 220110
PEX1	5189	AR	Heimler syndrome 1, 234580|Peroxisome biogenesis disorder 1A (Zellweger), 214100|Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	5192	AR	Peroxisome biogenesis disorder 6A (Zellweger), 614870|Peroxisome biogenesis disorder 6B, 614871
PEX11B	8799	AR	Peroxisome biogenesis disorder 14B, 614920
PEX12	5193	AR	Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	5194	AR	Peroxisome biogenesis disorder 11A (Zellweger), 614883
PEX16	9409	AR	Peroxisome biogenesis disorder 8A, (Zellweger), 614876
PEX19	5824	AR	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	5828	AR	Peroxisome biogenesis disorder 5A (Zellweger), 614866
PEX26	55670	AR	Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	8504	AR	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	5830	AR	Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	5190	AR	Heimler syndrome 2, 616617
PEX7	5191	AR	Peroxisome biogenesis disorder 9B, 614879
PGAP1	80055	AR	Mental retardation, autosomal recessive 42, 615802
PGAP2	27315	AR	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	93210	AR	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	5230	XLR	Phosphoglycerate kinase 1 deficiency, 300653
PGM3	5238	AR	Immunodeficiency 23, 615816
PHACTR1	221692	AD	OMIM618298
PHF21A	51317		POTOCKI-SHAFFER SYNDROME, 601224
PHF6	84295	XLR	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	23133	XLR	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	26227	AR	Neu-Laxova syndrome 1, 256520
PHIP	55023	AD	Intellectual disability (de Ligt (2012) N Engl J Med 367,1921)|Glaucoma,primary congenital (Lee (2011) Mol Vis 17,3583)
PIDD1	55367	AR	PMID: 29302074
PIGA	5277	XL	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	9488	AR	PMID:31256876; OMIM #618580
PIGC	5279	AR	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	54872	AR	Mental retardation,autosomal recessive 53,616917
PIGK	10026	AR	PMID:32220290
PIGL	9487	AR	CHIME syndrome, 280000
PIGN	23556	AR	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	84720	AR	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGQ	9091	AR	OMIM618548
PIGS	94005	AR	PMID: 30269814
PIGT	51604	AR	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	128869	AR	PMID:31353022; OMIM #618590
PIGV	55650	AR	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	284098	AR	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	84992	AR	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	5290	AD	Breast cancer, somatic, 114480|Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501
PIK3R2	5296	AD	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PLA2G6	8398	AR	Infantile neuroaxonal dystrophy 1, 256600|Neurodegeneration with brain iron accumulation 2B, 610217|Parkinson disease 14, autosomal recessive, 612953
PLAA	9373	AR	Neurodevelopmental disorder with progressive microcephaly,

			spasticity, and brain anomalies, 617527
PLCB1	23236	AR	Epileptic encephalopathy, early infantile, 12, 613722
PLK4	10733	AR	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	5354	XLR	Pelizaeus-Merzbacher disease, 312080|Spastic paraplegia 2, X-linked, 312920
PLPBP	11212	AR	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLXNA3	55558	AR	PMID:31130284
PMM2	5373	AR	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	23203	AR	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	9512	AR	PMID: 29576218
PNKP	11284	AR	Ataxia-oculomotor apraxia 4, 616267|Microcephaly, seizures, and developmental delay, 613402
PNP	4860	AR	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	10908	AR	?Laurence-Moon syndrome, 245800
PNPO	55163	AR	OMIM610090
POGZ	23126	AD	White-Sutton syndrome, 616364
POLA1	5422	XLR	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, OMIM:310465(Intellectual disability|Spasticity)|OMIM:301220(Intellectual disability|Global developmental delay)
POLG	5428	AR	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700
POLR1D	51082	AD/AR	OMIM613717
POLR2A	5430	AD	PMID: 31353023; OMIM #618603
POLR3A	11128	AR	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	55703	AR	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POLRMT	5442	AD/AR	PMID: 33602924
POMGNT1	55624	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280|Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151
POMGNT2	84892	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	84197	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 615249
POMT1	10585	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670|Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	29954	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150|Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
PORCN	64840	XLD	Focal dermal hypoplasia, 305600
POU1F1	5449	AD	Pituitary hormone deficiency, combined, 1, 613038
POU3F2	5454	AD	Intellectual Disability and Dysmorphic Features Dheedene et al Mol Syndromol 2014;
POU3F3	5455	AD	DD, ID, speech/language delay
PPM1D	8493	AD	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold 617450
PPOX	5498	AD	Porphyria variegata, 176200
PPP1CB	5500	AD	Intellectual disability, syndromic (Ma (2016) Hum Genet 135, 1399)
PPP1R12A	4659	AD	PMID:31883643
PPP1R15B	84919	AR	Microcephaly, short stature, and impaired glucose metabolism 2, 616817

PPP1R21	129285	AR	PMID: 29808498
PPP2CA	5515	AD	PMID:30595372; OMIM #618354
PPP2R1A	5518	AD	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	5528	AD	Mental retardation, autosomal dominant 35, 616355
PPP3CA	5530	AD	AD.Epileptic encephalopathy infantile or early childhood 1 .#617711
PPT1	5538	AR	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	10084	XLR	Renpenning syndrome, 309500
PRICKLE1	144165	AR	OMIM612437
PRKACA	5566	AD	PMID:33058759
PRKACB	5567	AD	PMID:33058759
PRKAR1A	5573	AD	https://doi.org/10.1101/797787; PMID 30006632
PRKD1	5587	AD	Congenital heart defects and ectodermal dysplasia 617364, OMIM:617364(Delayed speech and language development|Global developmental delay)
PRMT7	54496	AR	PMID:26437029; OMIM #617157
PRODH	5625	AR	Hyperprolinemia, type I, 239500|{Schizophrenia, susceptibility to, 4}, 600850
PRPS1	5631	XLR	Arts syndrome, 301835|Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070
PRR12	57479	AD	PMID: 29556724
PRRT2	112476	AD	BENIGN FAMILIAL INFANTILE EPILEPSY AND INFANTILE CONVULSIONS WITH CHOREOATHETOSIS SYNDROME, OMIM:128200(Paroxysmal choreoathetosis)
PRSS12	8492	AR	Mental retardation, autosomal recessive 1, 249500
PRUNE1	58497	AR	PMID: 30556349; OMIM #617481
PSAP	5660	AR	Combined SAP deficiency, 611721|Gaucher disease, atypical, 610539|Krabbe disease, atypical, 611722
PSMD12	5718	AD	Stankiewicz-Isidor syndrome, 617516;
PSPH	5723	AR	Phosphoserine phosphatase deficiency, 614023
PTCH1	5727	AD	Basal cell carcinoma, somatic, 605462|Holoprosencephaly-7, 610828
PTCHD1	139411	XL	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	9791	AD	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	5728	AD	Bannayan-Riley-Ruvalcaba syndrome, 153480|Macrocephaly/autism syndrome, 605309
PTF1A	256297	AR	Pancreatic agenesis 2, 615935
PTPN11	5781	AD	LEOPARD syndrome 1, 151100|Noonan syndrome 1, 163950
PTPN12	5782	AR	PMID:31130284
PTRH2	51651	AR	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	391356	AR	PMID: 30940925
PTS	5805	AR	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	22827	AD	Verheij syndrome, 615583
PUM1	9698	AD	PMID: 29474920
PURA	5813	AD	Mental retardation, autosomal dominant 31, 616158
PUS1	80324	AR	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	83480	AR	Mental retardation, autosomal recessive 55, 617051
PUS7	54517	AR	PMID: 30526862
PYCR1	5831	AR	Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	29920	AR	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	5859	AR	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	5860	AR	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	54870	AD	Ververi-Brady syndrome 617982
RAB11B	9230	AD	Neurodevelopmental disorder with ataxic gait absent speech and decreased cortical white matter . #617807
RAB18	22931	AR	Warburg micro syndrome 3, 614222

RAB23	51715	AR	Carpenter syndrome, 201000
RAB27A	5873	AR	Griscelli syndrome, type 2, 607624
RAB39B	116442	XL	Mental retardation, X-linked 72, 300271
RAB3GAP1	22930	AR	Warburg micro syndrome 1, 600118
RAB3GAP2	25782	AR	Martsolf syndrome, 212720|Warburg micro syndrome 2, 614225
RAB40AL	282808	XL	?Martin-Probst syndrome (Bedoyan (2012) J Med Genet 49, 332)
RAC1	5879	AD	Mental retardation, autosomal dominant 48 617751
RAC3	5881	AD	PMID: 30293988
RAD21	5885	AD	Cornelia de Lange syndrome 4, 614701
RAF1	5894	AD	Cardiomyopathy, dilated, 1NN, 615916|Noonan syndrome 5, 611553|LEOPARD syndrome 2, 611554
RAI1	10743	AD	Smith-Magenis syndrome, 182290
RALA	5898	AD	PMID: 30500825
RALGAPA1	253959	AR	PMID:32004447
RALGAPB	57148	AD	Autism,Guo et al., Mol Psy 2018
RARB	5915	AD	https://doi.org/10.1101/797787; PMID 27120018
RARS2	57038	AR	Pontocerebellar hypoplasia, type 6, 611523
RASIP1	54922	AR	PMID:31130284
RBBP8	5932	AR	Jawad syndrome, 251255
RBM10	8241	XLR	TARP syndrome, 311900
RBM28	55131	AR	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	3516	AD	OMIM614814
RCBTB1	55213	AR	[Beta-glycopyranoside tasting]
RECQL4	9401	AR	Baller-Gerold syndrome, 218600
RELN	5649	AR	Lissencephaly 2 (Norman-Roberts type), 257320
RERE	473	AD	Neurodevelopmental disorder with or without anomalies of the brain,eye or heart, 616975
RFT1	91869	AR	Congenital disorder of glycosylation, type In, 612015
RHOBTB2	23221	AD	Epileptic encephalopathy, early infantile, 64 618004 AD
RIMS2	9699	AR	PMID:32470375
RIT1	6016	AD	Noonan syndrome 8, 615355
RLIM	51132	XLR	Mental Retardation, X-linked 61, 300978
RMND1	55005	AR	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	6023	AR	Anauxetic dysplasia 1, 607095
RNASEH2A	10535	AR	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	79621	AR	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	84153	AR	Aicardi-Goutieres syndrome 3, 610329
RNASET2	8635	AR	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	7737	XLD	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	54941	AD	Tenorio syndrome, 616260
RNF13	11342	AD	PMID:30595371; OMIM #618379
ROGDI	79641	AR	Kohlschutter-Tonz syndrome, 226750
ROR2	4920	AD/AR	Brachydactyly, type B1, 113000
RORA	6095	AD	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia 618060 AD
RORB	6096	AD	OMIM618357
RPGRIP1L	23322	AR	COACH syndrome, 216360|Joubert syndrome 7, 611560|Meckel syndrome 5, 611561
RPL10	6134	XL	{Autism, susceptibility to, X-linked 5}, 300847
RPS6KA3	6197	XLD	Coffin-Lowry syndrome, 303600|Mental retardation, X-linked 19, 300844
RRAS2	22800	AD	PMID:31130285
RRM2B	50484	AR	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075
RSPRY1	89970	AR	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	51319	AR	Intellectual developmental disorder, autosomal recessive 70,

			618402
RTEL1	51750	AR	Dyskeratosis congenita, autosomal dominant 4, 615190
RTN4IP1	84816	AR	OMIM #616732
RTTN	25914	AR	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	9711	AR	SYNDROMIC MR WITH ATAXIA, DYSARTHRIA AND EPILEPSY, OMIM:615705(Motor delay|Cerebellar atrophy|Intellectual disability)
RUSC2	9853	AR	Mental retardation, autosomal recessive 61, 617773
RXYLT1	10329	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SALL1	6299	AD	Townes-Brocks branchiootorenal-like syndrome, 107480|Townes-Brocks syndrome, 107480
SAMD9	54809	AD	MIRAGE syndrome, 617053
SAMHD1	25939	AR	?Chilblain lupus 2, 614415
SATB2	23314	AD	Glass syndrome, 612313
SBDS	51119	AR	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SC5D	6309	AR	Lathosterolosis, 607330
SCAF4	57466	AD	PMID:32730804
SCAMP5	192683	AD	PMID:31439720
SCAPER	49855	AR	AR retinitis pigmentosa with ID
SCARB2	950	AR	OMIM254900
SCN1A	6323	AD	Dravet syndrome, 607208|Epilepsy, generalized, with febrile seizures plus, type 2, 604403
SCN1B	6324	AR	Atrial fibrillation, familial, 13, 615377|Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	6326	AD	Epileptic encephalopathy, early infantile, 11, 613721|Seizures, benign familial infantile, 3, 607745
SCN3A	6328	AD	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	6334	AD	Epileptic encephalopathy, early infantile, 13, 614558|?Cognitive impairment with or without cerebellar ataxia, 614306
SCO1	6341	AR	Mitochondrial complex IV deficiency, 220110
SCO2	9997	AR	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCYL1	57410	AR	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	10806	AR	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	6389	AR	Cardiomyopathy, dilated, 1GG, 613642|Leigh syndrome, 256000
SEMA3E	9723	AD	?CHARGE syndrome, 214800
SEMA6B	10501	AD	PMID:32169168
SEPSECS	51091	AR	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	84947	AR	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINI1	5274	AR	OMIM604218
SET	6418	AD	Intellectual disability;SET syndrome (GenomicsEngland,
SETBP1	26040	AD	Mental retardation, autosomal dominant 29, 616078|Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	9739	AD	PMID: 26974950
SETD1B	23067	AD	PMID: 31440728
SETD2	29072	AD	Luscan-Lumish syndrome, 616831
SETD5	55209	AD	Mental retardation, autosomal dominant 23, 615761
SGPL1	8879	AR	Nephrotic syndrome, type 14, 617575
SGSH	6448	AR	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SHANK2	22941	AD	{Autism susceptibility 17}, 613436
SHANK3	85358	AD	Phelan-McDermid syndrome, 606232
SHH	6469	AD	Holoprosencephaly-3, 142945|Schizencephaly, 269160
SHOC2	8036	AD	Noonan-like syndrome with loose anagen hair, 607721

SHROOM4	57477	XL	?Stocco dos Santos X-linked mental retardation syndrome, 300434
SIK1	150094	AD	Epileptic encephalopathy, early infantile, 30 616341 AD
SIL1	64374	AR	Marinesco-Sjogren syndrome, 248800
SIN3A	25942	AD	Witteveen-Kolk syndrome, 613406
SIX3	6496	AD	Holoprosencephaly-2, 157170|Schizencephaly, 269160
SKI	6497	AD	Shprintzen-Goldberg syndrome, 182212
SLC12A5	57468	AD	Epileptic encephalopathy, early infantile, 34, 616645
SLC12A6	9990	AR	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	284111	AR	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A2	6567	XLD	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	26503	AR	Salla disease, 604369
SLC19A3	80704	AR	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	6505	AR	Dicarboxylic aminoaciduria, 222730|{?Schizophrenia susceptibility 18}, 615232
SLC1A2	6506	AD	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	6509	AR	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A12	8604	AR	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	10166	AR	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	79751	AR	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	29957	AD	OMIM612289
SLC2A1	6513	AD	Dystonia 9, 601042
SLC33A1	9197	AD	Congenital cataracts, hearing loss, and neurodegeneration, 614482|Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	10559	AR	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	7355	XLD	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	23443	AR	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	55343	AR	Congenital disorder of glycosylation, type IIc, 266265
SLC39A14	23516	AR	?Hyperostosis cranalis interna, 144755
SLC39A8	64116	AR	Congenital disorder of glycosylation, type IIn 616721 AR
SLC46A1	113235	AR	Folate malabsorption, hereditary, 229050
SLC4A4	8671	AR	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A1	6529	AD	Myoclonic-atonic epilepsy, 616421
SLC6A17	388662	AR	Mental retardation, autosomal recessive 48, 616269
SLC6A19	340024	AR	Hartnup disorder, 234500
SLC6A3	6531	AR	Parkinsonism-dystonia, infantile, 613135
SLC6A8	6535	XLR	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	6536	AR	AR.Glycine encephalopathy with normal serum glycine.#617301
SLC7A5	8140	AR	PMID:27912058
SLC7A7	9056	AR	Lysinuric protein intolerance, 222700
SLC9A6	10479	XLD	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	84679	AD	PMID: 30335141
SMAD4	4089	AD	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050|Myhre syndrome, 139210
SMAD6	4091	AD	Lelieveld et al Nature Neuroscience (2016)(ID)
SMARCA2	6595	AD	Nicolaides-Baraitser syndrome, 601358
SMARCA4	6597	AD	Coffin-Siris syndrome 4, 614609
SMARCB1	6598	AD	Coffin-Siris syndrome 3, 614608|?Ivemark syndrome (Carss (2014) Hum Mol Genet 23,3269)|?Autism (Neale (2012) Nature 485,242)
SMARCC2	6601	AD	PMID:30580808
SMARCD1	6602	AD	PMID: 30879640

SMARCD2	6603	AR	Specific granule deficiency 2 617475 (includes global developmental delay in some patients)
SMARCE1	6605	AD	Coffin-Siris syndrome 5,616938
SMC1A	8243	XLR	Cornelia de Lange syndrome 2,300590
SMC3	9126	AD	Cornelia de Lange syndrome 3, 610759
SMG8	55181	AR	PMID: 31130284
SMG9	56006	AR	Heart and brain malformation syndrome, OMIM:616920(Global brain atrophy|Cerebral atrophy|Global developmental delay)
SMO	6608		Curry-Jones syndrome, somatic mosaic 601707, OMIM:601707(Global developmental delay|Agenesis of corpus callosum)
SMOC1	64093	AR	Microphthalmia with limb anomalies, 206920
SMPD1	6609	AR	Niemann-Pick disease, type A, 257200|Niemann-Pick disease, type B, 607616
SMPD4	55627	AR	OMIM618622 PMID:31130284
SMS	6611	XLR	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	6616	AD	PMID: 29942082
SNAP29	9342	AR	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	79753	AR	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRPB	6628	AD	Cerebrocostomandibular syndrome 117650 AD
SNRPN	6638	AD	Prader-Willi syndrome, 176270
SNX14	57231	AR	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	55084	AR	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SON	6651	AD	ZITK syndrome, 617140
SOS1	6654	AD	Noonan syndrome 4, 610733
SOS2	6655	AD	Noonan syndrome 9, 616559
SOX10	6663	AD	PCWH syndrome, 609136|Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	6664	AD	Mental retardation, autosomal dominant, 27, 615866
SOX2	6657	AD	Microphthalmia, syndromic 3, 206900|Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	6658	XL	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123|Panhypopituitarism, X-linked, 312000
SOX4	6659	AD	PMID: 30661772; OMIM #618506
SOX5	6660	AD	Lamb-Shaffer syndrome, 616803
SOX6	55553	AD	PMID:32442410,
SOX9	6662	AD	OMIM114290
SPART	23111	AR	Troyer syndrome, 275900
SPAST	6683	AD	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	166378	AR	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPECC1L	23384	AD	OPITZ GBBB SYNDROME, TYPE II 145410 AD
SPEN	23013	AD	PMID: 33596411
SPG11	80208	AR	Amyotrophic lateral sclerosis 5, juvenile, 602099|Charcot-Marie-Tooth disease, axonal, type 2X, 616668|Spastic paraplegia 11, autosomal recessive, 604360
SPOP	8405	AD	PMID:32109420
SPR	6697	AR	PMID: 11443547
SPRED1	161742	AD	Legius syndrome, 611431
SPTAN1	6709	AD	Epileptic encephalopathy, early infantile, 5, 613477
SPTBN2	6712	AR	Spinocerebellar ataxia 5, 600224
SPTBN4	57731	AR	PMID: 29861105
SPTLC2	9517	AD	Neuropathy hereditary sensory and autonomic type IC 61364
SQSTM1	8878	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, OMIM:617145(Cerebellar atrophy|Abnormal pyramidal signs)|OMIM:616437(Cerebral cortical atrophy|Language impairment)

SRCAP	10847	AD	Floating-Harbor syndrome, 136140
SRD5A3	79644	AR	Congenital disorder of glycosylation, type Iq, 612379|Kahrizi syndrome, 612713
SREBF1	6720	AD	PMID:32497488
SRGAP3	9901		3p- syndrome MIM:613792 (includes intellectual disability)
SRPX2	27286	AD	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SSR4	6748	XL	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	6487	AR	Epileptic encephalopathy, early infantile, 15, 615006|Mental retardation, autosomal recessive 12, 611090
ST3GAL5	8869	AR	Amish infantile epilepsy syndrome, 609056
STAG1	10274	AD	?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)|?Schizophrenia (Li (2016) Mol Psychiatry 21,290)
STAG2	10735	XD	Mullegama-Klein-Martinez syndrome 301022 XL
STAMBP	10617	AR	Microcephaly-capillary malformation syndrome, 614261
STARD9	57519	AD	PMID: 28777490
STIL	6491	AR	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	64220	AR	Microphthalmia, isolated, with coloboma 8, 601186|Microphthalmia, syndromic 9, 601186
STRADA	92335	AR	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STT3A	3703	AR	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	201595	AR	?Congenital disorder of glycosylation, type Ix, 615597
STX1B	112755	AD	OMIM616172
STXBP1	6812	AD	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	8803	AR	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	8802	AR	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	51684	AD/AR	Basal cell nevus syndrome| Joubert syndrome, OMIM:109400|617757(Motor delay|Intellectual disability)
SUMF1	285362	AR	Multiple sulfatase deficiency, 272200
SUOX	6821	AR	Sulfite oxidase deficiency, 272300
SUPT16H	11198	AD	PMID:31924697
SURF1	6834	AR	Charcot-Marie-Tooth disease, type 4K, 616684|Leigh syndrome, due to COX IV deficiency, 256000
SUZ12	23512		No OMIM phenotype, PMID: 30019515
SVBP	374969	AR	PMID:30607023; OMIM #618569
SYN1	6853	XLD	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	10492	AD	?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)
SYNE1	23345	AR	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 |Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNGAP1	8831	AD	Mental retardation, autosomal dominant 5, 612621
SYNJ1	8867	AR	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	6855	XLD	Mental retardation, X-linked 96, 300802
SYT1	6857	AD	INTELLECTUAL DISABILITY
SYT14	255928	AR	Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	23334	AR	Epileptic encephalopathy, early infantile, 18, 615476
TAF1	6872	XLR	Dystonia-Parkinsonism, X-linked, 314250|Mental retardation, X-linked, syndromic 33, 300966
TAF13	6884	AR	Mental retardation autosomal recessive 60 OMIM:617432
TAF2	6873	AR	Mental retardation, autosomal recessive 40, 615599
TAF6	6878	AR	PMID:29302074
TANC2	26115	AD	PMID:31616000
TANGO2	128989	AR	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration,
TAOK1	57551	AD	PMID:31230721

TASP1	55617	AR	PMID:31209944
TAT	6898	AR	Tyrosinemia, type II, 276600
TBC1D20	128637	AR	Warburg micro syndrome 4, 615663
TBC1D23	55773	AR	AR.Pontocerebellar hypoplasia type 11.# 617695
TBC1D24	57465	AD/AR	Deafness , autosomal recessive 86, 614617|Epileptic encephalopathy, early infantile, 16, 615338
TBC1D7	51256	AR	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	6904	AR	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	6905	AR	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410|Kenny-Caffey syndrome, type 1, 244460
TBCK	93627	AR	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3,616900
TBL1XR1	79718	AD/AR	Mental retardation, autosomal dominant 41,616944|Piermont syndrome,602342
TBR1	10716	AD	Intellectual disability (Hamdan (2014) PLoS Genet 10)
TBX1	6899	AD	Conotruncal anomaly face syndrome, 217095
TCF20	6942	AD	PMID: 31238879
TCF4	6925	AD	Corneal dystrophy, Fuchs endothelial, 3, 613267|Pitt-Hopkins syndrome, 610954
TCN2	6948	AR	Transcobalamin II deficiency, 275350
TCTN2	79867	AR	?Meckel syndrome 8, 613885
TCTN3	26123	AR	Joubert syndrome 18, 614815
TDP2	51567	AR	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	9895	AR	Spastic paraplegia 49, autosomal recessive, 615031
TECR	9524	AR	Mental retardation, autosomal recessive 14, 614020
TELO2	9894	AR	You-Hoover-Fong syndrome, 616954
TET3	200424	AD/AR	PMID:31928709
TFAP2A	7020	AD	Branchiooculofacial syndrome, 113620
TGFBR1	7046	AD	Loeys-Dietz syndrome 1, 609192|{Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGIF1	7050	AD	Holoprosencephaly-4, 142946
TH	7054	AR	Segawa syndrome, recessive, 605407
THOC2	57187	XLR	Mental retardation, X-linked 12/35, 300957
THOC6	79228	AR	Beaulieu-Boycott-Innes syndrome, 613680
THRB	7068	AD	Thyroid hormone resistance, 188570
TIMM50	92609	AR	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	1678	XLR	Jensen syndrome, 311150
TINF2	26277	AD	Revesz syndrome 268130 AD
TK2	7084	AR	PMID: 25446393
TKFC	26007	AR	PMID:32004446
TKT	7086	AR	Short stature, developmental delay, and congenital heart defects, OMIM:617044(Intellectual disability)
TLK2	11011	AD	Lelieveld et al Nature Neuroscience (2016)(ID)
TMCO1	54499	AR	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM165	55858	AR	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	51259	AR	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	79583	AR	Joubert syndrome 20, 614970
TMEM237	65062	AR	Joubert syndrome 14, 614424
TMEM240	339453	AD	Spinocerebellar ataxia 21, 607454
TMEM67	91147	AR	COACH syndrome, 216360|Joubert syndrome 6, 610688 |Meckel syndrome 3, 607361|Nephronophthisis 11, 613550
TMEM70	54968	AR	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	9772	AR	Intellectual developmental disorder with cardiac defects and dysmorphic facies 618316 (KIAA0195)
TMLHE	55217	XL	{Autism,susceptibility to,X-linked 6}, 300872

TMTC3	160418	AR	Lissencephaly 8, 617255 (includes intellectual disability), OMIM:617255(Intellectual disability|Occipital encephalocele|Global developmental delay)
TMX2	51075	AR	PMID:31586943
TNIK	23043	AR	Mental retardation, autosomal recessive 54, 617028
TNRC6B	23112	AD	PMID:32152250
TOE1	114034	AR	Pontocerebellar hypoplasia, type 7, 614969
TP73	7161	AR	PMID:31130284
TPP1	1200	AR	Ceroid lipofuscinosis, neuronal, 2, 204500
TPP2	7174	AR	OMIM619220 PMID: 33586135
TRAF7	84231	AD	PMID: 29961569; OMIM #618164
TRAIP	10293	AR	Seckel syndrome 9, 616777
TRAK1	22906	AR	OMIM618201
TRAPPC11	60684	AR	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC12	51112	AR	AR.Encephalopathy progressive early-onset with brain atrophy and spasticity.#617669
TRAPPC6B	122553	AR	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	83696	AR	Mental retardation, autosomal recessive 13, 613192
TREX1	11277	AD/AR	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750|Chilblain lupus, 610448
TRIM32	22954	AR	?Bardet-Biedl syndrome 11, 615988
TRIO	7204	AD	Mental retardation, autosomal dominant 44,617061
TRIP12	9320	AD	Mental retardation, autosomal dominant 49 617752
TRIT1	54802	AR	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	55621	AR	Mental retardation, autosomal recessive 68 618302
TRMT10A	93587	AR	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRPM3	80036	AD	PMID:31278393
TRRAP	8295	AD	PMID:30827496; OMIM #618454
TSC1	7248	AD	Lymphangioleiomyomatosis, 606690|Tuberous sclerosis-1, 191100
TSC2	7249	AD	Lymphangioleiomyomatosis, somatic, 606690|Tuberous sclerosis-2, 613254
TSEN15	116461	AR	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	80746	AR	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4, OMIM:612389(Cerebellar hypoplasia|Cerebral atrophy|Progressive microcephaly)
TSEN34	79042	AR	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4, OMIM:612390(Cerebellar vermis hypoplasia|Cerebellar hemisphere hypoplasia)
TSEN54	283989	AR	Pontocerebellar hypoplasia type 2A, 277470|Pontocerebellar hypoplasia type 4, 225753
TSFM	10102	AR	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	7252	AR	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	7102	XLR	Mental retardation, X-linked 58, 300210
TTC19	54902	AR	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	9652	AR	Trichohepatoenteric syndrome 1, 222470
TTC5	91875	AR	PMID: 29302074
TTC8	123016	AR	Bardet-Biedl syndrome 8, 615985
TTI2	80185	AR	Mental retardation, autosomal recessive 39, 615541
TUBA1A	7846	AD	Lissencephaly 3, 611603
TUBA8	51807	AR	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB	203068	AD	Cortical dysplasia, complex, with other brain malformations 6 615771 AD
TUBB2A	7280	AD	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	347733	AR	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	10381	AD	Cortical dysplasia, complex, with other brain malformations 1

			614039 AD
TUBB4A	10382	AD	OMIM612438
TUBG1	7283	AD	Cortical dysplasia, complex, with other brain malformations 4 615412 AD
TUBGCP2	10844	AR	PMID:31630790
TUBGCP4	27229	AR	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	85378	AR	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	7991	AR	Mental retardation, autosomal recessive 7, 611093
TWIST1	7291	AD	Craniosynostosis, type 1, 123100|Robinow-Sorauf syndrome, 180750|Saethre-Chotzen syndrome, 101400
UBA5	79876	AR	Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	7319	XLR	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	7337	AD	Angelman syndrome, 105830
UBE3B	89910	AR	Kaufman oculocerebrofacial syndrome, 244450
UBR1	197131	AR	Johanson-Blizzard syndrome, 243800
UBTF	7343	AD	developmental regression;motor and language regression;developmental delay;Neurodegeneration childhood-onset with brain atrophy 617672
UFC1	51506	AR	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	51569	AR	Leukodystrophy, hypomyelinating, 14, 617899
UGDH	7358	AR	OMIM618792
UNC80	285175	AR	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	51733	AR	Beta-ureidopropionase deficiency, 613161
UPF3B	65109	XLR	Mental retardation, X-linked, syndromic 14, 300676
UROC1	131669	AR	?Urocanase deficiency, 276880
USP27X	389856	XLR	Intellectual disability (Hu (2015) Mol Psychiatry epub,epub)
USP7	7874	AD	PMID: 26365382
USP9X	8239	XLD/XLR	Mental retardation, X-linked 99, 300919|Mental retardation, X-linked 99, syndromic, female-restricted, 300968
VAC14	55697	AR	Striatonigral degeneration, childhood-onset, OMIM:617054(Delayed speech and language development|Developmental regression)
VAMP1	6843	AR/AD	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VAMP2	6844	AD	PMID:30929742
VARS1	7407	AR	OMIM617802
VARS2	57176	AR	OMIM615917
VLDLR	7436	AR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	55823	AR	Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	157680	AR	Cohen syndrome, 216550
VPS37A	137492	AR	Spastic paraplegia 53, autosomal recessive, 614898
VPS4A	27183	AD/AR	PMID:33186543
VPS53	55275	AR	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	7443	AR	Pontocerebellar hypoplasia type 1A, 607596
VWA3B	200403	AR	?Spinocerebellar ataxia, autosomal recessive 22, 616948
WAC	51322	AD	Desanto-Shinawi syndrome, 616708
WARS2	10352	AR	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	8936	AD	PMID: 29961568
WASHC4	23325	AR	?Mental retardation, autosomal recessive 43, OMIM:615817(Intellectual disability|Spasticity|Global developmental delay)
WBP11	51729	AD	PMID:33276377

WDFY3	23001	AD	?Microcephaly 18, primary, autosomal dominant 617520
WDPCP	51057	AR	?Bardet-Biedl syndrome 15, 615992
WDR13	64743	XL	Intellectual disability,X-linked (Whibley (2010) Am J Hum Genet 87,173)
WDR19	57728	AR	Nephronophthisis 13, 614377|Senior-Loken syndrome 8, 616307
WDR26	80232	AD	Skraban-Deardorff syndrome 617616
WDR37	22884	AD	PMID:31327508; OMIM #618652
WDR4	10785	AR	Galloway-Mowat syndrome 6, 618347
WDR45	11152	XLD	Neurodegeneration with brain iron accululation 5, 300894
WDR45B	56270	AR	AUTOSOMAL RECESSIVE MENTAL RETARDATION (GenomicsEngland,
WDR62	284403	AR	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	84942	AR	Galloway-Mowat syndrome 1 251300 AR
WDR81	124997	AR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	51741	AR	Epileptic encephalopathy, early infantile, 28, 616211|Spinocrebellar ataxia, autosomal recessive 12, 614322
XPA	7507	AR	Xeroderma pigmentosum, group A, 278700
XRCC4	7518	AR	OMIM616541
XYLT1	64131	AR	Desbuquois dysplasia 2, 615777
YAP1	10413	AD	Coloboma, ocular, 120433|Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YWHAE	7531	AD	Develomental delay,facial dysmorphology and growth retardation (Enomoto (2012) Am J Med Genet A 158A)|Developmental delay and mild brain structural abnormalities (Bi (2009) Nat Genet 41,168)
YWHAG	7532	AD	# 617665. EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 56; EIEE56
YY1	7528	AD	Mental retardation (Vissers (2010) Nat Genet 42,1109)
ZBTB11	27107	AR	PMID: 29893856
ZBTB16	7704	AR	Leukemia, acute promyelocytic, PL2F/RARA type|Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	10472	AD	?Mental retardation, autosomal dominant 22, 612337
ZBTB20	26137	AD	Primrose syndrome 259050 AD
ZBTB24	9841	AR	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZC3H14	79882	AR	Mental retardation, autosomal recessive 56, 617125
ZC4H2	55906	XL	Wieacker-Wolff syndrome, 314580
ZDHHC9	51114	XL	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	9839	AD	Mowat-Wilson syndrome, 235730
ZFYVE26	23503	AR	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	7545	AD	Craniosynostosis 6 616602 AD
ZIC2	7546	AD	Holoprosencephaly-5, 609637
ZMIZ1	57178	AD	PMID:30639322; OMIM #618659
ZMYND11	10771	AD	Mental retardation, autosomal dominant 30, 616083
ZNF142	7701	AR	PMID:31036918; OMIM #618425
ZNF148	7707	AD	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies 617260 AD
ZNF292	23036	AD	?Autism (Neale (2012) Nature 485,242)|Autism,Guo et al., Mol Psy 2018
ZNF407	55628	AR	PMID: 24907849
ZNF41	7592	XL	Mental retardation, X-linked 89, 300848
ZNF462	58499	AD	PMID: 31361404, PMID: 28513610
ZNF526	116115	AR	Non-syndromic autosomal recessive intellectual disability (Marangi et al, Eur J Hum Genet. 2013
ZNF711	7552	XL	Mental retardation, X-linked 97, 300803
ZNHIT3	9326	AR	OMIM260565

ZSWIM6	57688	AD	Acromelic frontonasal dysostosis, 603671