ID and epilepsy

Gene panel

Gene panel information

Gene panel	**ID and epilepsy**
Version	9
Total genes	1969
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Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AAAS	99.88 %	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AARS1	99.99 %	601065	Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive
AARS2	99.98 %	612035	Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive
AASS	99.61 %	605113	Hyperlysinemia, 238700 (3), Autosomal recessive
ABAT	99.98 %	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCA2	100 %	600047	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive
ABCC8	99.98 %	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal dominant, Autosomal recessive; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal dominant, Autosomal recessive
ABCC9	99.92 %	601439	Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
ABCD1	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABCD4	100 %	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
ABHD16A	100 %	142620	Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive
ABHD5	99.98 %	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ACACA	99.92 %	200350	Acetyl-CoA carboxylase deficiency, 613933 (3), Autosomal recessive
ACAD9	100 %	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACADM	96.14 %	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
ACADS	99.99 %	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
ACADSB	99.93 %	600301	2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive
ACAT1	99.81 %	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
ACBD6	98.86 %	616352	Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive
ACER3	99.76 %	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive

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ACO2	99.99 %	100850	Optic atrophy 9, 616289 (3), Autosomal dominant, Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
ACOX1	99.98 %	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACSF3	99.99 %	614245	Combined malonic and methylmalonic aciduria, 614265 (3), Autosomal recessive
ACSL4	99.59 %	300157	Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant
ACTB	100 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479 (3)
ACTG1	100 %	102560	Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant
ACTL6A	99.74 %	604958	*No OMIM phenotypes*
ACTL6B	99.9 %	612458	Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant
ACVR1	99.94 %	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACY1	100 %	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
ADAM22	99.15 %	603709	Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive
ADAR	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADARB1	94.29 %	601218	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive
ADAT3	100 %	615302	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive
ADCY5	99.98 %	600293	Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive
ADD3	99.95 %	601568	Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive
ADGRG1	99.9 %	604110	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752 (3); Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854 (3), Autosomal recessive
ADGRL1	99.98 %	616416	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065 (3), Autosomal dominant
ADGRL2	98.18 %	607018	*No OMIM phenotypes*
ADK	99.78 %	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
ADNP	100 %	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
ADPRS	99.94 %	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
ADSL	99.93 %	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
AFF2	99.89 %	300806	Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive
AFF3	99.63 %	601464	KINSSHIP syndrome, 619297 (3), Autosomal dominant
AFF4	99.94 %	604417	CHOPS syndrome, 616368 (3), Autosomal dominant

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AFG3L2	99.97 %	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGA	99.92 %	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGMO	99.77 %	613738	*No OMIM phenotypes*
AGO1	99.73 %	606228	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292 (3), Autosomal dominant
AGO2	99.97 %	606229	Lessel-Kreienkamp syndrome, 619149 (3), Autosomal dominant
AGPAT2	100 %	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGTPBP1	99.68 %	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
AHCY	100 %	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
AHDC1	100 %	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
AHI1	99.86 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AIFM1	99.92 %	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
AIMP1	99.97 %	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
AIMP2	99.99 %	600859	Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive
AK1	100 %	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
AKT3	99.81 %	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant
ALDH18A1	99.96 %	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
ALDH3A2	99.95 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH4A1	98.97 %	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
ALDH5A1	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALDH7A1	99.49 %	107323	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100 (3), Autosomal recessive
ALG1	86.66 %	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG11	99.99 %	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
ALG12	100 %	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
ALG13	99.44 %	300776	Developmental and epileptic encephalopathy 36, 300884 (3), X-linked
ALG14	99.34 %	612866	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3), Autosomal recessive; Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive
ALG2	100 %	607905	Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
ALG3	99.96 %	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
ALG6	93.37 %	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
ALG8	95.49 %	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant

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ALG9	99.73 %	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type II, 608776 (3), Autosomal recessive
ALKBH8	99.98 %	613306	Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive
ALMS1	99.9 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALPL	99.88 %	171760	Odontohypophosphatasia, 146300 (3), Autosomal dominant, Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal dominant, Autosomal recessive
ALX1	97.99 %	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
ALX4	100 %	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
AMACR	100 %	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
AMER1	100 %	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
AMFR	99.74 %	603243	Spastic paraplegia 89, autosomal recessive, 620379 (3), Autosomal recessive
AMMECR1	99.87 %	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
AMOTL1	99.96 %	614657	*No OMIM phenotypes*
AMPD2	99.91 %	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, autosomal recessive, 615686 (3), Autosomal recessive
AMT	100 %	238310	Glycine encephalopathy 2, 620398 (3)
ANK2	99.98 %	106410	Long QT syndrome 4, 600919 (3), Autosomal dominant; Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant
ANK3	99.79 %	600465	Intellectual developmental disorder, autosomal recessive 37, 615493 (3), Autosomal recessive
ANKLE2	99.99 %	616062	Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
ANKRD11	99.85 %	611192	KBG syndrome, 148050 (3), Autosomal dominant
ANKRD17	99.92 %	615929	Chopra-Amiel-Gordon syndrome, 619504 (3), Autosomal dominant
ANKS1B	99.74 %	607815	*No OMIM phenotypes*
ANO10	99.93 %	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
ANO4	99.78 %	610111	*No OMIM phenotypes*
ANTXR1	99.96 %	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
AP1B1	99.99 %	600157	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive
AP1G1	99.95 %	603533	Usmani-Riazuddin syndrome, autosomal recessive, 619548 (3), Autosomal recessive; Usmani-Riazuddin syndrome, autosomal dominant, 619467 (3), Autosomal dominant
AP1S1	99.49 %	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP1S2	99.56 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
AP2M1	99.69 %	601024	Intellectual developmental disorder 60 with seizures, 618587 (3), Autosomal dominant
AP2S1	99.98 %	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant
AP3B1	99.89 %	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3B2	100 %	602166	Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive
AP3D1	100 %	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
AP4B1	96.92 %	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive

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Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AP4E1	99.94 %	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
AP4M1	99.98 %	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
AP4S1	87.89 %	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
AP5Z1	100 %	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
APC2	99.98 %	612034	Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 74, 617169 (3), Autosomal recessive
APTX	99.92 %	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
ARCN1	99.92 %	600820	Short stature-micrognathia syndrome, 617164 (3), Autosomal dominant
ARF1	99.99 %	103180	Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant
ARF3	99.99 %	103190	*No OMIM phenotypes*
ARFGEF1	99.86 %	604141	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964 (3), Autosomal dominant
ARFGEF2	99.99 %	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
ARG1	99.95 %	608313	Argininemia, 207800 (3), Autosomal recessive
ARHGAP31	100 %	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARHGEF9	99.89 %	300429	Developmental and epileptic encephalopathy 8, 300607 (3), X-linked
ARID1A	99.83 %	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	99.69 %	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARID2	99.48 %	609539	Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant
ARL13B	99.53 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6	99.9 %	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
ARMC9	99.77 %	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
ARPC4	100 %	604226	Developmental delay, language impairment, and ocular abnormalities, 620141 (3), Autosomal dominant
ARSA	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSL	99.93 %	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ARV1	99.85 %	611647	Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive
ARX	95.36 %	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
ASAH1	99.9 %	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASCC3	99.76 %	614217	Intellectual developmental disorder, autosomal recessive 81, 620700 (3), Autosomal recessive
ASH1L	99.79 %	607999	Intellectual developmental disorder, autosomal dominant 52, 617796 (3), Autosomal dominant
ASL	99.98 %	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASNS	99.63 %	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
ASPA	99.98 %	608034	Canavan disease, 271900 (3), Autosomal recessive
ASPM	99.57 %	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ASS1	77.52 %	603470	Citrullinemia, 215700 (3), Autosomal recessive
ASXL1	100 %	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
ASXL2	99.82 %	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ASXL3	99.99 %	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant
ATAD1	99.85 %	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
ATAD3A	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATCAY	100 %	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATG4D	99.97 %	611340	*No OMIM phenotypes*
ATG7	99.9 %	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive
ATIC	99.86 %	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATL1	99.95 %	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
ATM	99.83 %	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATN1	99.9 %	607462	Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant
ATP13A2	99.96 %	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
ATP1A1	98.41 %	182310	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	99.98 %	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
ATP2A2	99.98 %	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP2B1	99.39 %	108731	Intellectual developmental disorder, autosomal dominant 66, 619910 (3), Autosomal dominant
ATP2B2	99.99 %	108733	Deafness, autosomal dominant 82, 619804 (3), Autosomal dominant; {Deafness, autosomal recessive 12, modifier of}, 601386 (3), Autosomal recessive
ATP5F1A	99.99 %	164360	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358 (3), Autosomal dominant; ?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive; ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228 (3), Autosomal recessive
ATP5F1E	100 %	606153	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3), Autosomal recessive
ATP5PO	99.94 %	600828	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 (3), Autosomal recessive

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ATP6AP2	99.55 %	300556	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive
ATP6V0A1	99.85 %	192130	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant
ATP6V0A2	99.92 %	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
ATP6V0C	100 %	108745	Epilepsy, early-onset, 3, with or without developmental delay, 620465 (3), Autosomal dominant
ATP6V1A	99.73 %	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant
ATP6V1B2	99.99 %	606939	Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant
ATP7A	99.87 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
ATP8A2	100 %	605870	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATP9A	99.99 %	609126	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242 (3), Autosomal recessive
ATPAF2	99.96 %	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
ATR	99.83 %	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
ATRX	99.44 %	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive; Alpha-thalassemia/impaired intellectual development syndrome, 301040 (3), X-linked dominant
ATXN2	99.88 %	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
ATXN7L3	99.97 %	619010	*No OMIM phenotypes*
AUH	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
AUTS2	99.87 %	607270	Intellectual developmental disorder, autosomal dominant 26, 615834 (3), Autosomal dominant
AVPR2	100 %	300538	Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
B3GALNT2	92.79 %	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive
B3GALT6	100 %	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive
B3GLCT	99.9 %	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALNT1	99.97 %	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
B4GALT1	99.97 %	137060	Combined low LDL and fibrinogen, 620364 (3), Autosomal recessive; Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive
B4GALT7	99.99 %	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
B4GAT1	100 %	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
B9D1	99.8 %	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	99.88 %	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
BAP1	99.99 %	603089	Kury-Isidor syndrome, 619762 (3), Autosomal dominant; Tumor predisposition syndrome 1, 614327 (3), Autosomal dominant; {Uveal melanoma, susceptibility to, 2}, 606661 (3), Autosomal dominant
BAZ2B	99.63 %	605683	*No OMIM phenotypes*
BBS1	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	99.98 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	100 %	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	99.9 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BBS4	99.88 %	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	99 %	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	99.42 %	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	99.75 %	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCAP31	99.95 %	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCAS3	99.31 %	607470	Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive
BCKDHA	99.97 %	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	99.73 %	248611	Maple syrup urine disease, type Ib, 620698 (3), Autosomal recessive
BCKDK	99.99 %	614901	Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3), Autosomal recessive
BCL11A	99.93 %	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant
BCL11B	100 %	606558	Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant
BCOR	99.97 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BCORL1	99.99 %	300688	Shukla-Vernon syndrome, 301029 (3), X-linked recessive
BCS1L	99.99 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
BICD2	99.99 %	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
BICRA	99.99 %	605690	Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant
BLM	99.8 %	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLOC1S1	99.95 %	601444	*No OMIM phenotypes*
BMP4	100 %	112262	Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BOLA3	99.22 %	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
BORCS8	99.93 %	616601	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
BPTF	99.84 %	601819	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant; {Kaposi sarcoma, susceptibility to}, 148000 (3), Autosomal dominant
BRAF	99.78 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
BRAT1	100 %	614506	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
BRD4	99.98 %	608749	Cornelia de Lange syndrome 6, 620568 (3), Autosomal dominant
BRF1	100 %	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
BRF2	100 %	607013	*No OMIM phenotypes*
BRPF1	100 %	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
BRSK2	99.98 %	609236	*No OMIM phenotypes*
BRWD3	99.4 %	300553	Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive
BSCL2	99.99 %	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
BTD	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
BUB1	99.64 %	602452	Colorectal cancer with chromosomal instability, somatic, 114500 (3); Microcephaly 30, primary, autosomal recessive, 620183 (3), Autosomal recessive
BUB1B	100 %	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
C12orf4	99.93 %	616082	Intellectual developmental disorder, autosomal recessive 66, 618221 (3), Autosomal recessive
C12orf57	100 %	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
C2CD3	99.88 %	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
C2orf69	99.97 %	619219	Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive
CA2	99.62 %	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CA5A	99.99 %	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
CA8	99.71 %	114815	Spinocerebellar ataxia, autosomal recessive 34, 613227 (3), Autosomal recessive
CACNA1A	98.16 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1B	100 %	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CACNA1C	100 %	114205	Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3), Autosomal dominant
CACNA1D	99.98 %	114206	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive
CACNA1E	99.82 %	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
CACNA1G	99.95 %	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
CACNA1I	99.98 %	608230	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114 (3), Autosomal dominant
CACNA2D1	97.12 %	114204	Developmental and epileptic encephalopathy 110, 620149 (3), Autosomal recessive
CACNA2D2	99.99 %	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
CACNB4	99.2 %	601949	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; ?Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant
CACNG2	99.99 %	602911	?Intellectual developmental disorder, autosomal dominant 10, 614256 (3), Autosomal dominant
CAD	99.86 %	114010	Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive
CAMK2A	99.99 %	114078	Intellectual developmental disorder, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 63, 618095 (3), Autosomal recessive
CAMK2B	99.92 %	607707	Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant
CAMK2D	99.71 %	607708	*No OMIM phenotypes*
CAMK2G	99.95 %	602123	Intellectual developmental disorder, autosomal dominant 59, 618522 (3), Autosomal dominant
CAMK4	99.78 %	114080	*No OMIM phenotypes*
CAMSAP1	99.99 %	613774	Cortical dysplasia, complex, with other brain malformations 12, 620316 (3), Autosomal recessive
CAMTA1	99.97 %	611501	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant
CAPN10	99.99 %	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
CAPN15	99.98 %	603267	Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive
CAPRIN1	99.56 %	601178	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782 (3), Autosomal dominant; Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636 (3), Autosomal dominant
CARNMT1	99.93 %	616552	*No OMIM phenotypes*
CARS1	99.99 %	123859	Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive
CARS2	99.99 %	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
CASK	98.95 %	300172	Intellectual developmental disorder, with or without nystagmus, 300422 (3), X-linked recessive; Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked; FG syndrome 4, 300422 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CASP2	99.99 %	600639	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653 (3), Autosomal recessive
CBL	99.95 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CBS	17.79 %	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
CC2D1A	99.98 %	610055	Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive
CC2D2A	99.95 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCBE1	99.52 %	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC115	99.9 %	613734	Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive
CCDC174	99.98 %	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
CCDC186	99.88 %	619249	*No OMIM phenotypes*
CCDC22	99.91 %	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
CCDC32	99.97 %	618941	Cardiofacioneurodevelopmental syndrome, 619123 (3), Autosomal recessive
CCDC47	99.97 %	618260	Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive
CCDC88A	99.48 %	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
CCDC88C	100 %	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
CCM2	99.93 %	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
CCND2	100 %	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
CCNK	99.93 %	603544	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant
CDC42	98.05 %	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
CDC42BPB	99.99 %	614062	Chilton-Okur-Chung neurodevelopmental syndrome, 619841 (3), Autosomal dominant
CDH11	99.94 %	600023	Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elsahy-Waters syndrome, 211380 (3), Autosomal recessive
CDH15	99.98 %	114019	Intellectual developmental disorder, autosomal dominant 3, 612580 (3), Autosomal dominant
CDH2	99.84 %	114020	Arrhythmogenic right ventricular dysplasia 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant
CDK10	99.98 %	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
CDK13	99.83 %	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
CDK19	99.26 %	614720	Developmental and epileptic encephalopathy 87, 618916 (3), Autosomal dominant
CDK5RAP2	99.97 %	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDK6	99.47 %	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
CDK8	99.81 %	603184	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant
CDKL5	99.88 %	300203	Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CDKN1C	100 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CELF2	99.99 %	602538	Developmental and epileptic encephalopathy 97, 619561 (3), Autosomal dominant
CELSR3	99.99 %	604264	*No OMIM phenotypes*
CENPF	99.97 %	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CENPJ	99.92 %	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
CEP104	99.99 %	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
CEP120	99.9 %	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
CEP135	99.82 %	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
CEP152	99.93 %	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
CEP290	98.1 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CEP295	99.94 %	617728	Seckel syndrome 11, 620767 (3), Autosomal recessive
CEP41	99.99 %	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP55	99.92 %	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
CEP57	99.92 %	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
CEP63	94.73 %	614724	?Seckel syndrome 6, 614728 (3), Autosomal recessive
CEP83	98.68 %	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CEP85L	100 %	618865	Lissencephaly 10, 618873 (3), Autosomal dominant
CERS1	100 %	606919	Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive
CERT1	99.66 %	604677	Intellectual developmental disorder, autosomal dominant 34, 616351 (3), Autosomal dominant
CHAMP1	99.99 %	616327	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579 (3), Autosomal dominant
CHD1	99.46 %	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
CHD2	99.97 %	602119	Developmental and epileptic encephalopathy 94, 615369 (3), Autosomal dominant
CHD3	99.06 %	602120	Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant
CHD4	99.99 %	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD5	99.97 %	610771	Parenti-Mignot neurodevelopmental syndrome, 619873 (3), Autosomal dominant
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHD8	99.96 %	610528	Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant
CHKA	99.95 %	118491	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023 (3), Autosomal recessive
CHKB	100 %	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHMP1A	100 %	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
CHRNA2	99.98 %	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
CHRNA4	100 %	118504	{Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CHRNB2	99.99 %	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CIC	98.32 %	612082	Intellectual developmental disorder, autosomal dominant 45, 617600 (3), Autosomal dominant
CIT	99.99 %	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
CKAP2L	99.54 %	616174	Filippi syndrome, 272440 (3), Autosomal recessive
CLCN3	99.95 %	600580	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 (3), Autosomal recessive; Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 (3), Autosomal dominant
CLCN4	99.98 %	302910	Raynaud-Claes syndrome, 300114 (3), X-linked dominant
CLCN6	100 %	602726	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173 (3), Autosomal dominant
CLCNKB	99.98 %	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN11	100 %	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
CLDN16	99.98 %	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	99.02 %	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLDN5	100 %	602101	*No OMIM phenotypes*
CLIC2	99.82 %	300138	*No OMIM phenotypes*
CLN3	99.92 %	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	100 %	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
CLN8	100 %	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive
CLP1	99.98 %	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
CLPB	99.97 %	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
CLTC	99.56 %	118955	Intellectual developmental disorder, autosomal dominant 56, 617854 (3), Autosomal dominant
CNKSR2	99.54 %	300724	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked
CNNM2	99.94 %	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 (3), Autosomal dominant, Autosomal recessive
CNOT1	99.83 %	604917	Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant
CNOT2	99 %	604909	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant
CNOT3	99.99 %	604910	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant
CNOT9	90.78 %	612054	*No OMIM phenotypes*
CNPY3	99.98 %	610774	Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive
CNTN2	99.95 %	190197	Epilepsy, early-onset, 5, with or without developmental delay, 615400 (3), Autosomal recessive
CNTNAP1	99.98 %	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CNTNAP2	99.99 %	604569	Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3)
COA8	99.94 %	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
COASY	99.98 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COG1	100 %	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
COG4	99.96 %	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
COG5	99.92 %	606821	Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive
COG6	99.86 %	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
COG7	99.74 %	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
COG8	100 %	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
COL18A1	99.99 %	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
COL4A1	99.99 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COL4A2	99.98 %	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
COLEC11	100 %	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
COLGALT1	99.82 %	617531	Brain small vessel disease 3, 618360 (3), Autosomal recessive
COPB1	99.87 %	600959	Baralle-Macken syndrome, 619255 (3), Autosomal recessive
COPB2	99.83 %	606990	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive
COQ2	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ4	100 %	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive; Spastic ataxia 10, autosomal recessive, 620666 (3), Autosomal recessive
COQ5	99.95 %	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
COQ7	100 %	601683	Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 (3), Autosomal recessive
COQ8A	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ9	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	99.99 %	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
COX11	99.72 %	603648	Mitochondrial complex IV deficiency, nuclear type 23, 620275 (3), Autosomal recessive
COX15	100 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
COX6B1	100 %	124089	Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive
CPA6	99.99 %	609562	Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal dominant, Autosomal recessive
CPE	99.93 %	114855	BDV syndrome, 619326 (3), Autosomal recessive
CPLANE1	99.81 %	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CPLX1	100 %	605032	Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive
CPS1	99.91 %	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
CPSF3	99.93 %	606029	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 (3), Autosomal recessive
CPT2	99.65 %	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal dominant, Autosomal recessive; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal dominant, Autosomal recessive
CRADD	99.9 %	603454	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
CRB2	99.95 %	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
CRBN	99.97 %	609262	Intellectual developmental disorder, autosomal recessive 2, 607417 (3), Autosomal recessive
CREBBP	99.97 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRELD1	99.99 %	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; Jeffries-Lakhani neurodevelopmental syndrome, 620771 (3), Autosomal recessive; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
CRIPT	99.96 %	604594	Rothmund-Thomson syndrome, type 3, 615789 (3), Autosomal recessive
CRLF1	99.99 %	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CRMP1	100 %	602462	*No OMIM phenotypes*
CRPPA	99.98 %	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
CSDE1	97.17 %	191510	*No OMIM phenotypes*
CSF1R	99.92 %	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant
CSMD1	99.99 %	608397	*No OMIM phenotypes*
CSMD2	99.33 %	608398	*No OMIM phenotypes*
CSNK1G1	99.95 %	606274	*No OMIM phenotypes*
CSNK2A1	99.96 %	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
CSNK2B	99.65 %	115441	Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant
CSPP1	98.31 %	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CSTB	100 %	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CTBP1	99.98 %	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
CTC1	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTCF	99.87 %	604167	Intellectual developmental disorder, autosomal dominant 21, 615502 (3), Autosomal dominant
CTDP1	99.97 %	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CTNNA2	99.84 %	114025	Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive
CTNNB1	99.95 %	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3)
CTNND1	99.92 %	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
CTNND2	99.97 %	604275	*No OMIM phenotypes*
CTR9	99.98 %	609366	*No OMIM phenotypes*
CTSA	99.98 %	613111	Galactosialidosis, 256540 (3), Autosomal recessive
CTSD	100 %	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CTSF	99.96 %	603539	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive
CTTNBP2	99.97 %	609772	*No OMIM phenotypes*
CTU2	99.91 %	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive
CUBN	99.99 %	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
CUL3	99.76 %	603136	Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
CUL4B	99.67 %	300304	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive
CUX1	99.37 %	116896	Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant
CUX2	99.96 %	610648	Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant
CWC27	99.67 %	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
CWF19L1	99.91 %	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
CYB5R3	99.93 %	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
CYC1	99.86 %	123980	Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive
CYFIP2	100 %	606323	Developmental and epileptic encephalopathy 65, 618008 (3), Autosomal dominant
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP2U1	99.99 %	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
D2HGDH	100 %	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
DAG1	100 %	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive
DAGLA	99.93 %	614015	Neuroocular syndrome 2, paroxysmal type, 168885 (3), Autosomal dominant
DARS1	98.85 %	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
DARS2	98.31 %	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DBT	94.51 %	248610	Maple syrup urine disease, type II, 620699 (3), Autosomal recessive
DCAF17	99.84 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DCC	99.96 %	120470	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive
DCHS1	100 %	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
DCPS	99.98 %	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DCX	99.99 %	300121	Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked
DDB1	99.96 %	600045	White-Kernohan syndrome, 619426 (3), Autosomal dominant
DDC	99.67 %	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DDHD2	99.97 %	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDOST	99.93 %	602202	Congenital disorder of glycosylation, type Ir, 614507 (3), Autosomal recessive
DDX11	99.74 %	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
DDX17	99.97 %	608469	*No OMIM phenotypes*
DDX23	99.94 %	612172	*No OMIM phenotypes*
DDX3X	99.01 %	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
DDX59	99.67 %	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DDX6	99.88 %	600326	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant
DEAF1	99.9 %	602635	Vulto-van Silfout-de Vries syndrome, 615828 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 (3), Autosomal recessive
DEGS1	99.99 %	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DENND5A	99.99 %	617278	Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive
DENND5B	95.23 %	617279	*No OMIM phenotypes*
DEPDC5	99.18 %	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant; Developmental and epileptic encephalopathy 111, 620504 (3), Autosomal recessive
DHCR24	99.93 %	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHDDS	98.65 %	608172	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHFR	98.89 %	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DHPS	93.17 %	600944	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive
DHRSX	100 %	301034	*No OMIM phenotypes*
DHTKD1	99.95 %	614984	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive
DHX16	99.98 %	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
DHX30	99.96 %	616423	Neurodevelopmental disorder with variable motor and speech impairment, 617804 (3), Autosomal dominant
DHX37	99.98 %	617362	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46XY sex reversal 11, 273250 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DHX9	99.52 %	603115	*No OMIM phenotypes*
DIAPH1	99.95 %	602121	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DIP2B	99.65 %	611379	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630 (3), Autosomal dominant
DIP2C	100 %	611380	*No OMIM phenotypes*
DIS3L2	99.9 %	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DISP1	99.93 %	607502	*No OMIM phenotypes*
DKC1	99.59 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLAT	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DLG3	99.96 %	300189	Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive
DLG4	99.99 %	602887	Intellectual developmental disorder, autosomal dominant 62, 618793 (3), Autosomal dominant
DLG5	99.91 %	604090	Yuksel-Vogel-Bauser syndrome, 620703 (3), Autosomal recessive
DLL1	99.99 %	606582	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant
DMD	99.76 %	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DMXL2	99.86 %	612186	Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
DNAJC12	99.72 %	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
DNAJC19	99.76 %	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC5	99.99 %	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant
DNAJC6	99.48 %	608375	Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive
DNM1	92.28 %	602377	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352 (3), Autosomal recessive; Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346 (3), Autosomal dominant
DNM1L	99.4 %	603850	Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal dominant, Autosomal recessive
DNMT3A	100 %	602769	Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant
DNMT3B	99.98 %	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
DOCK3	99.96 %	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive
DOCK4	99.95 %	607679	*No OMIM phenotypes*
DOCK6	100 %	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK7	94.41 %	615730	Developmental and epileptic encephalopathy 23, 615859 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DOCK8	99.86 %	611432	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700 (3), Autosomal recessive
DOHH	100 %	611262	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066 (3), Autosomal recessive
DOLK	100 %	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DONSON	99.99 %	611428	Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive
DOT1L	99.98 %	607375	*No OMIM phenotypes*
DPAGT1	100 %	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive
DPF2	99.99 %	601671	Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant
DPH1	100 %	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive
DPH2	99.97 %	603456	Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062 (3), Autosomal recessive
DPH5	96.83 %	611075	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070 (3), Autosomal recessive
DPM1	90.68 %	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPM2	100 %	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
DPM3	99.98 %	605951	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive
DPP6	99.99 %	126141	Intellectual developmental disorder, autosomal dominant 33, 616311 (3), Autosomal dominant; {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
DPYD	94.53 %	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
DPYS	99.99 %	613326	Dihydropyrimidinuria, 222748 (3), Autosomal recessive
DPYSL5	99.93 %	608383	Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant
DRG1	99.95 %	603952	Tan-Almurshedi syndrome, 620641 (3), Autosomal recessive
DTYMK	99.99 %	188345	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 (3), Autosomal recessive
DYM	99.96 %	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
DYNC1H1	99.99 %	600112	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant
DYNC1I2	98.97 %	603331	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (3), Autosomal recessive
DYRK1A	99.98 %	600855	Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant
EARS2	99.96 %	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
EBF3	99.99 %	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
EBP	99.92 %	300205	MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ECHS1	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
ECM1	99.99 %	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
EDC3	99.97 %	609842	?Intellectual developmental disorder, autosomal recessive 50, 616460 (3), Autosomal recessive
EDEM3	98.38 %	610214	Congenital disorder of glycosylation, type IIv, 619493 (3), Autosomal recessive
EED	93.81 %	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant
EEF1A2	100 %	602959	Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 38, 616393 (3), Autosomal dominant
EEF1B2	99.59 %	600655	*No OMIM phenotypes*
EEF1D	99.99 %	130592	*No OMIM phenotypes*
EEF2	99.95 %	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
EFNB2	99.92 %	600527	*No OMIM phenotypes*
EFTUD2	99.93 %	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EHMT1	98.38 %	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
EIF2AK2	99.7 %	176871	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant; Dystonia 33, 619687 (3), Autosomal dominant, Autosomal recessive
EIF2AK3	97.43 %	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
EIF2B1	99.98 %	606686	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive
EIF2B2	99.9 %	606454	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3), Autosomal recessive
EIF2B3	97.26 %	606273	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3), Autosomal recessive
EIF2B4	99.96 %	606687	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3), Autosomal recessive
EIF2B5	99.98 %	603945	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3), Autosomal recessive
EIF2S3	99.34 %	300161	MEHMO syndrome, 300148 (3), X-linked recessive
EIF3F	99.96 %	603914	Intellectual developmental disorder, autosomal recessive 67, 618295 (3), Autosomal recessive
EIF4A2	99.99 %	601102	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455 (3), Autosomal dominant, Autosomal recessive
EIF4A3	99.99 %	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
EIF5A	100 %	600187	Faundes-Banka syndrome, 619376 (3), Autosomal dominant
ELAC2	99.9 %	605367	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive
ELFN1	100 %	614964	*No OMIM phenotypes*
ELOVL4	99.91 %	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 (3), Autosomal recessive
ELP2	99.82 %	616054	Intellectual developmental disorder, autosomal recessive 58, 617270 (3), Autosomal recessive
EMC1	99.85 %	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EMC10	99.96 %	614545	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 (3), Autosomal recessive
EML1	99.99 %	602033	Band heterotopia, 600348 (3), Autosomal recessive
EMX2	100 %	600035	Schizencephaly, 269160 (3)
ENTPD1	99.98 %	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
EP300	99.97 %	602700	Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EP400	99.96 %	606265	*No OMIM phenotypes*
EPB41L1	100 %	602879	?Intellectual developmental disorder, autosomal dominant 11, 614257 (3), Autosomal dominant
EPB41L3	99.98 %	605331	*No OMIM phenotypes*
EPG5	99.95 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPM2A	99.99 %	607566	Myoclonic epilepsy of Lafora 1, 254780 (3), Autosomal recessive
EPRS1	99.53 %	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
ERBB4	99.92 %	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant
ERCC1	99.96 %	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC2	99.98 %	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
ERCC3	99.9 %	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC5	99.99 %	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	99.6 %	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
ERCC6L2	99.94 %	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
ERCC8	99.79 %	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
ERI1	99.8 %	608739	Hoxha-Aliu syndrome, 620662 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663 (3), Autosomal recessive
ERLIN2	99.94 %	611605	Spastic paraplegia 18A, autosomal dominant, 620512 (3), Autosomal dominant; Spastic paraplegia 18B, autosomal recessive, 611225 (3), Autosomal recessive
ESAM	99.99 %	614281	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371 (3), Autosomal recessive
ESCO2	99.92 %	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
ETFA	99.88 %	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	100 %	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	99.82 %	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
ETHE1	84.97 %	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
EXOC2	99.97 %	615329	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive
EXOC3L2	99.88 %	616927	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EXOC7	100 %	608163	Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3), Autosomal recessive
EXOSC2	100 %	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
EXOSC3	100 %	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC5	99.98 %	606492	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 (3), Autosomal recessive
EXOSC8	99.91 %	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXOSC9	94.91 %	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
EXT2	99.98 %	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
EXTL3	99.99 %	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
EZH2	99.89 %	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FA2H	99.98 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAM111A	100 %	615292	Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant
FAM126A	99.81 %	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FAM149B1	99.69 %	618413	Joubert syndrome 36, 618763 (3), Autosomal recessive
FAM177A1	99.98 %	619181	*No OMIM phenotypes*
FAM20C	100 %	611061	Raine syndrome, 259775 (3), Autosomal recessive
FAM50A	99.99 %	300453	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261 (3), X-linked recessive
FANCD2	99.86 %	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FAR1	99.82 %	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
FARS2	100 %	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FARSA	100 %	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	99.64 %	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
FASTKD2	99.93 %	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
FAT4	99.98 %	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
FBRSL1	95.66 %	620123	*No OMIM phenotypes*
FBXL3	99.48 %	605653	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive
FBXL4	100 %	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
FBXO11	99.53 %	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant
FBXO28	99.67 %	609100	Developmental and epileptic encephalopathy 100, 619777 (3), Autosomal dominant
FBXO31	99.99 %	609102	?Intellectual developmental disorder, autosomal recessive 45, 615979 (3), Autosomal recessive
FBXW11	99.96 %	605651	Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FBXW7	99.9 %	606278	Developmental delay, hypotonia, and impaired language, 620012 (3), Autosomal dominant
FCSK	99.97 %	608675	Congenital disorder of glycosylation with defective fucosylation 2, 618324 (3), Autosomal recessive
FDFT1	99.99 %	184420	Squalene synthase deficiency, 618156 (3), Autosomal recessive
FEM1B	100 %	613539	*No OMIM phenotypes*
FGD1	99.96 %	300546	Intellectual developmental disorder, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive
FGF12	99.94 %	601513	Developmental and epileptic encephalopathy 47, 617166 (3), Autosomal dominant
FGF13	99.76 %	300070	Developmental and epileptic encephalopathy 90, 301058 (3), X-linked recessive, X-linked dominant; Intellectual developmental disorder, X-linked 110, 301095 (3), X-linked recessive
FGF14	99.99 %	601515	Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant
FGFR1	100 %	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR2	99.99 %	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant
FGFR3	100 %	134934	Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal dominant, Autosomal recessive; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); LADD syndrome 2, 620192 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant
FH	99.95 %	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive
FIBP	99.83 %	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FIG4	99.83 %	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive
FILIP1	99.98 %	607307	Neuromuscular disorder, congenital, with dysmorphic facies, 620775 (3), Autosomal recessive
FITM2	99.99 %	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FKRP	100 %	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive
FKTN	99.94 %	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive
FLNA	99.99 %	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FLVCR1	99.91 %	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FLVCR2	100 %	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
FMN2	99.99 %	606373	Intellectual developmental disorder, autosomal recessive 47, 616193 (3), Autosomal recessive
FMR1	99.56 %	309550	Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
FOLR1	100 %	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FOSL2	99.95 %	601575	Aplasia cutis-enamel dysplasia syndrome, 620789 (3), Autosomal dominant
FOXG1	99.91 %	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXP1	99.98 %	605515	Intellectual developmental disorder with language impairment with or without autistic features, 613670 (3), Autosomal dominant
FOXP2	99.98 %	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
FOXP4	99.98 %	608924	*No OMIM phenotypes*
FOXRED1	100 %	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
FRA10AC1	99.76 %	608866	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113 (3), Autosomal recessive
FRAS1	99.97 %	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
FREM2	99.97 %	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
FRMD4A	100 %	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMD5	100 %	616309	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 (3), Autosomal dominant
FRMPD4	99.92 %	300838	Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked
FRRS1L	99.91 %	604574	Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive
FRYL	99.78 %	620798	*No OMIM phenotypes*
FTCD	99.99 %	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
FTH1	22.62 %	134770	Neurodegeneration with brain iron accumulation 9, 620669 (3), Autosomal dominant; ?Hemochromatosis, type 5, 615517 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FTO	99.66 %	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
FTSJ1	99.95 %	300499	Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive
FUCA1	98.72 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUT8	99.98 %	602589	Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive
FXYD2	100 %	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
FZR1	99.99 %	603619	Developmental and epileptic encephalopathy 109, 620145 (3), Autosomal dominant
GABBR1	99.91 %	603540	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502 (3), Autosomal dominant
GABBR2	99.96 %	607340	{Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3); Developmental and epileptic encephalopathy 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant
GABRA1	100 %	137160	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3)
GABRA2	99.9 %	137140	Developmental and epileptic encephalopathy 78, 618557 (3), Autosomal dominant; {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial
GABRA3	99.96 %	305660	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091 (3), X-linked
GABRA5	99.96 %	137142	Developmental and epileptic encephalopathy 79, 618559 (3), Autosomal dominant
GABRB1	99.97 %	137190	Developmental and epileptic encephalopathy 45, 617153 (3), Autosomal dominant
GABRB2	99.92 %	600232	Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant
GABRB3	99.66 %	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Developmental and epileptic encephalopathy 43, 617113 (3), Autosomal dominant
GABRD	99.99 %	137163	{?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant
GABRG2	91.88 %	137164	Developmental and epileptic encephalopathy 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 3, 607681 (3), Autosomal dominant
GAD1	99.92 %	605363	Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive
GAL	99.9 %	137035	?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant
GALC	99.92 %	606890	Krabbe disease, 245200 (3), Autosomal recessive
GALE	99.9 %	606953	Thrombocytopenia 13, syndromic, 620776 (3), Autosomal recessive; Galactose epimerase deficiency, 230350 (3), Autosomal recessive
GALNT2	99.44 %	602274	Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive
GALT	100 %	606999	Galactosemia, 230400 (3), Autosomal recessive
GAMT	100 %	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GATAD2B	99.44 %	614998	GAND syndrome, 615074 (3), Autosomal dominant
GATM	99.92 %	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
GBA	96.92 %	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GCDH	100 %	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCK	99.99 %	138079	MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant
GCSH	98.75 %	238330	Multiple mitochondrial dysfunctions syndrome 7, 620423 (3), Autosomal recessive
GDI1	99.99 %	300104	Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant
GEMIN4	100 %	606969	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive
GEMIN5	99.96 %	607005	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive
GFAP	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
GFER	100 %	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
GFM1	99.95 %	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GFM2	99.87 %	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GIGYF1	99.98 %	612064	*No OMIM phenotypes*
GJB1	100 %	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
GJC2	100 %	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
GK	99.27 %	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
GLB1	100 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLDC	99.99 %	238300	Glycine encephalopathy1, 605899 (3), Autosomal recessive
GLI2	99.93 %	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	100 %	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLIS3	99.99 %	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
GLRA1	100 %	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
GLRA2	99.87 %	305990	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076 (3), X-linked
GLRB	99.79 %	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLS	99.78 %	138280	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive
GLUD1	99.8 %	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
GLUL	99.88 %	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive; Developmental and epileptic encephalopathy 116, 620806 (3), Autosomal dominant
GLYCTK	100 %	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GM2A	100 %	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMNN	99.73 %	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
GMPPA	99.97 %	615495	Alacrima, achalasia, and impaired intellectual development syndrome, 615510 (3), Autosomal recessive
GMPPB	100 %	615320	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive
GNAI1	99.7 %	139310	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854 (3), Autosomal dominant
GNAO1	99.86 %	139311	Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNAQ	99.94 %	600998	Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3)
GNAS	100 %	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GNB1	100 %	139380	Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant
GNB2	99.99 %	139390	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 (3), Autosomal dominant; ?Sick sinus syndrome 4, 619464 (3), Autosomal dominant
GNB5	99.98 %	604447	Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173 (3), Autosomal recessive
GNE	99.99 %	603824	Sialuria, 269921 (3), Autosomal dominant; Thrombocytopenia 12 with or without myopathy, 620757 (3), Autosomal recessive; Nonaka myopathy, 605820 (3), Autosomal recessive
GNPAT	99.78 %	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPTAB	99.76 %	607840	Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive
GNPTG	100 %	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNS	99.59 %	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GOLGA2	100 %	602580	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240 (3), Autosomal recessive
GOSR2	98.92 %	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive
GOT2	99.78 %	138150	Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive
GPAA1	100 %	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
GPC3	99.6 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GPC4	99.89 %	300168	Keipert syndrome, 301026 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GPHN	99.94 %	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
GPRC5B	99.81 %	605948	Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447 (3), Autosomal dominant
GPSM2	96.11 %	609245	Chudley-McCullough syndrome, 604213 (3), Autosomal recessive
GPT2	99.97 %	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
GRIA1	99.97 %	138248	?Intellectual developmental disorder, autosomal recessive 76, 619931 (3), Autosomal recessive; Intellectual developmental disorder, autosomal dominant 67, 619927 (3), Autosomal dominant
GRIA2	99.99 %	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant
GRIA3	99.93 %	305915	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive
GRIA4	99.93 %	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant
GRID2	99.97 %	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRIK2	99.9 %	138244	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 (3), Autosomal dominant; Intellectual developmental disorder, autosomal recessive 6, 611092 (3), Autosomal recessive
GRIN1	100 %	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GRIN2A	100 %	138253	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 (3), Autosomal dominant
GRIN2B	99.99 %	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
GRIN2D	99.97 %	602717	Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant
GRIP1	99.83 %	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GRM1	100 %	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
GRM7	99.99 %	604101	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive
GRN	100 %	138945	Frontotemporal dementia 2, 607485 (3), Autosomal dominant, Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant, Autosomal recessive; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
GSS	99.99 %	601002	Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive
GSX2	100 %	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
GTF2E2	100 %	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
GTF2H5	100 %	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
GTF3C3	99.86 %	604888	*No OMIM phenotypes*
GTF3C5	99.99 %	604890	*No OMIM phenotypes*
GTPBP2	99.98 %	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
GTPBP3	99.99 %	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GUSB	95.07 %	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
H1-4	100 %	142220	Rahman syndrome, 617537 (3), Autosomal dominant
H3-3A	35.67 %	601128	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 (3), Autosomal dominant
H3-3B	100 %	601058	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 (3), Autosomal dominant
H4C11	99.08 %	602826	?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759 (3), Autosomal dominant
H4C3	99.99 %	602827	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758 (3), Autosomal dominant
H4C5	100 %	602830	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950 (3), Autosomal dominant
H4C9	100 %	602833	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951 (3), Autosomal dominant
HAAO	99.96 %	604521	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive
HACE1	99.76 %	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
HADH	99.86 %	601609	Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive
HADHA	99.98 %	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency 1, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
HAX1	100 %	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HCCS	99.9 %	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HCFC1	99.99 %	300019	Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive
HCN1	99.99 %	602780	Developmental and epileptic encephalopathy 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant
HCN2	97.35 %	602781	Febrile seizures, familial, 2, 602477 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 11, 602477 (3), Autosomal dominant
HDAC3	100 %	605166	*No OMIM phenotypes*
HDAC4	99.98 %	605314	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 (3), Autosomal dominant
HDAC6	99.96 %	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
HDAC8	99.74 %	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HEATR3	99.72 %	614951	Diamond-Blackfan anemia 21, 620072 (3), Autosomal recessive
HECTD4	99.96 %	620209	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive
HECW2	99.91 %	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
HEPACAM	100 %	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 (3), Autosomal dominant
HERC1	99.92 %	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HERC2	95.66 %	605837	Intellectual developmental disorder, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
HESX1	99.77 %	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal dominant, Autosomal recessive; Septooptic dysplasia, 182230 (3), Autosomal dominant, Autosomal recessive; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal dominant, Autosomal recessive
HEXA	99.99 %	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HGSNAT	99.93 %	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HHAT	99.93 %	605743	Nivelon-Nivelon-Mabille syndrome, 600092 (3), Autosomal recessive
HIBCH	99.7 %	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HID1	99.99 %	605752	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 (3), Autosomal recessive
HIVEP2	99.99 %	143054	Intellectual developmental disorder, autosomal dominant 43, 616977 (3), Autosomal dominant
HK1	99.97 %	142600	Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive
HLCS	99.97 %	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMGB1	54.18 %	163905	*No OMIM phenotypes*
HMGCL	99.31 %	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HNMT	99.53 %	605238	Intellectual developmental disorder, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
HNRNPC	100 %	164020	Intellectual developmental disorder, autosomal dominant 74, 620688 (3), Autosomal dominant
HNRNPH1	99.98 %	601035	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083 (3), Autosomal dominant
HNRNPH2	99.96 %	300610	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant
HNRNPK	99.93 %	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HNRNPR	98.05 %	607201	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073 (3), Autosomal dominant
HNRNPU	99.95 %	602869	Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant
HOXA1	100 %	142955	Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive
HPD	99.99 %	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPDL	99.99 %	618994	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive
HPRT1	97.8 %	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HRAS	100 %	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
HS2ST1	95.79 %	604844	Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive
HSD17B10	99.98 %	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HSD17B4	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSPA9	99.96 %	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
HSPD1	83.42 %	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
HTRA2	99.99 %	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
HUWE1	99.88 %	300697	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked
HYAL2	100 %	603551	*No OMIM phenotypes*
IARS1	99.89 %	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IARS2	99.77 %	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
IBA57	100 %	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
IDH2	100 %	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDS	99.82 %	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	99.99 %	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
IER3IP1	99.92 %	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFIH1	99.84 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFT172	99.98 %	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	100 %	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT74	99.71 %	608040	Bardet-Biedl syndrome 22, 617119 (3), Autosomal recessive; Spermatogenic failure 58, 619585 (3), Autosomal recessive; Joubert syndrome 40, 619582 (3), Autosomal recessive
IGBP1	99.9 %	300139	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
IGF1	100 %	147440	Insulin-like growth factor I deficiency, 608747 (3), Autosomal recessive
IGF1R	100 %	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal dominant, Autosomal recessive
IKBKG	57.34 %	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IL1RAPL1	99.77 %	300206	Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive
IMPA1	99.95 %	602064	Intellectual developmental disorder, autosomal recessive 59, 617323 (3), Autosomal recessive
IMPDH2	100 %	146691	[IMPDH2 enzyme activity, variation in], 617995 (3)
INPP4A	99.78 %	600916	*No OMIM phenotypes*
INPP5E	99.85 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INPP5K	99.94 %	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
INTS1	100 %	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive
INTS11	100 %	611354	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 (3), Autosomal recessive
IQSEC1	99.99 %	610166	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive
IQSEC2	99.97 %	300522	Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant
IREB2	99.9 %	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
IRF2BPL	99.21 %	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
IRX5	100 %	606195	Hamamy syndrome, 611174 (3), Autosomal recessive
ISCA1	99.79 %	611006	Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive
ISCA2	100 %	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
ITGA7	99.87 %	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
ITPA	99.99 %	147520	[Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive
ITPR1	99.98 %	147265	Gillespie syndrome, 206700 (3), Autosomal dominant, Autosomal recessive; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant
ITSN1	99.89 %	602442	*No OMIM phenotypes*
IVD	100 %	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
JAG1	100 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAM2	91.82 %	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
JAM3	100 %	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JARID2	99.97 %	601594	Developmental delay with variable intellectual disability and dysmorphic facies, 620098 (3), Autosomal dominant
KANSL1	99.85 %	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KARS1	99.98 %	601421	Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive
KAT5	100 %	601409	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3), Autosomal dominant
KAT6A	99.93 %	601408	Arboleda-Tham syndrome, 616268 (3), Autosomal dominant
KAT6B	99.79 %	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
KAT8	99.98 %	609912	Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant
KATNAL2	99.98 %	614697	*No OMIM phenotypes*
KATNB1	99.99 %	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
KATNIP	99.13 %	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
KCNA1	100 %	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA2	99.99 %	176262	Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant
KCNA3	99.76 %	176263	*No OMIM phenotypes*
KCNA4	100 %	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCNB1	100 %	600397	Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant
KCNB2	100 %	607738	*No OMIM phenotypes*
KCNC1	100 %	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
KCNC2	99.88 %	176256	Developmental and epileptic encephalopathy 103, 619913 (3), Autosomal dominant
KCNC3	99.98 %	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
KCND1	99.99 %	300281	*No OMIM phenotypes*
KCND2	99.9 %	605410	*No OMIM phenotypes*
KCND3	99.98 %	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant
KCNH1	99.92 %	603305	Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant
KCNH5	99.89 %	605716	Developmental and epileptic encephalopathy 112, 620537 (3), Autosomal dominant
KCNJ10	99.98 %	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ11	100 %	600937	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal dominant, Autosomal recessive
KCNJ6	100 %	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNK3	100 %	603220	Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant
KCNK4	99.99 %	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant
KCNK9	100 %	605874	Birk-Barel syndrome, 612292 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KCNMA1	99.89 %	600150	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant
KCNN2	91.25 %	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
KCNN3	99.97 %	602983	Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant
KCNQ2	100 %	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCNQ3	99.98 %	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
KCNQ5	99.89 %	607357	Intellectual developmental disorder, autosomal dominant 46, 617601 (3), Autosomal dominant
KCNT1	99.98 %	608167	Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant
KCNT2	98.77 %	610044	Developmental and epileptic encephalopathy 57, 617771 (3), Autosomal dominant
KCTD3	99.75 %	613272	*No OMIM phenotypes*
KCTD7	99.98 %	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KDM1A	89.42 %	609132	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
KDM2B	99.99 %	609078	*No OMIM phenotypes*
KDM3B	99.98 %	609373	Diets-Jongmans syndrome, 618846 (3), Autosomal dominant
KDM4B	99.99 %	609765	Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant
KDM5A	99.9 %	180202	El Hayek-Chahrour neurodevelopmental syndrome, 620820 (3), Autosomal recessive
KDM5B	97.15 %	605393	Intellectual developmental disorder, autosomal recessive 65, 618109 (3), Autosomal recessive
KDM5C	99.98 %	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM6A	99.74 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KDM6B	99.99 %	611577	Stolerman neurodevelopmental syndrome, 618505 (3), Autosomal dominant
KIAA0586	95.75 %	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
KIAA1109	99.79 %	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
KICS2	99.94 %	617420	*No OMIM phenotypes*
KIDINS220	99.94 %	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogryposis, 619501 (3), Autosomal recessive
KIF11	99.84 %	148760	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 (3), Autosomal dominant
KIF14	97.8 %	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KIF1A	99.96 %	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant; Spastic paraplegia 30, autosomal recessive, 620607 (3), Autosomal recessive
KIF21B	99.83 %	608322	*No OMIM phenotypes*
KIF26A	100 %	613231	Cortical dysplasia, complex, with other brain malformations 11, 620156 (3), Autosomal recessive
KIF2A	99.79 %	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
KIF4A	99.76 %	300521	Taurodontism, microdontia, and dens invaginatus, 313490 (3), X-linked recessive; Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive
KIF5A	99.91 %	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
KIF5C	99.93 %	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
KIF7	100 %	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KIFBP	99.91 %	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
KIRREL3	99.99 %	607761	*No OMIM phenotypes*
KLF7	99.99 %	604865	*No OMIM phenotypes*
KLHL15	100 %	300980	Intellectual developmental disorder, X-linked 103, 300982 (3), X-linked recessive
KLHL20	99.51 %	617679	*No OMIM phenotypes*
KLHL7	99.95 %	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
KMT2A	99.97 %	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2B	99.99 %	606834	Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
KMT2C	98.98 %	606833	Kleefstra syndrome 2, 617768 (3), Autosomal dominant
KMT2D	99.98 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant
KMT2E	99.92 %	608444	O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant
KMT5B	99.91 %	610881	Intellectual developmental disorder, autosomal dominant 51, 617788 (3), Autosomal dominant
KNL1	98.43 %	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
KPTN	99.98 %	615620	Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive
KRAS	99.13 %	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
L1CAM	99.98 %	308840	MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus, congenital, X-linked, 307000 (3), X-linked recessive; ?Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive
L2HGDH	99.92 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAGE3	99.99 %	300060	Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive
LAMA1	99.98 %	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMA2	99.95 %	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
LAMB1	99.87 %	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
LAMC3	99.97 %	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
LAMP2	98.95 %	309060	Danon disease, 300257 (3), X-linked dominant
LARGE1	100 %	603590	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
LARP7	99.63 %	612026	Alazami syndrome, 615071 (3), Autosomal recessive
LARS1	99.87 %	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
LAS1L	99.97 %	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LETM1	99.97 %	604407	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive
LGI1	99.99 %	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
LGI3	100 %	608302	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007 (3), Autosomal recessive
LGI4	99.98 %	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
LHX2	100 %	603759	*No OMIM phenotypes*
LIAS	99.98 %	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
LIG3	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
LIG4	100 %	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LINGO1	100 %	609791	Intellectual developmental disorder, autosomal recessive 64, 618103 (3), Autosomal recessive
LINS1	99.96 %	610350	Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive
LIPT1	99.89 %	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
LMAN2L	99.91 %	609552	?Intellectual developmental disorder, autosomal dominant 69, 617863 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 52, 616887 (3), Autosomal recessive
LMBRD1	99.67 %	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive
LMBRD2	99.66 %	619490	Developmental delay with variable neurologic and brain abnormalities, 619694 (3), Autosomal dominant
LMNB1	99.73 %	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant
LMNB2	99.99 %	150341	Microcephaly 27, primary, autosomal dominant, 619180 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LNPK	92.81 %	610236	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive
LONP1	99.99 %	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LRP2	99.86 %	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRPPRC	99.8 %	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive
LRRC7	94.76 %	614453	*No OMIM phenotypes*
LSM7	99.99 %	607287	*No OMIM phenotypes*
LSS	99.98 %	600909	Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive
LYRM7	99.98 %	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LZTFL1	100 %	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
LZTR1	99.46 %	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
MAB21L1	100 %	601280	Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive
MAB21L2	100 %	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal dominant, Autosomal recessive
MACF1	99.33 %	608271	Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant
MADD	99.95 %	603584	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive
MAF	99.73 %	177075	Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant
MAG	99.99 %	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
MAGEL2	99.99 %	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
MAGT1	99.54 %	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive
MAN1B1	99.94 %	604346	Rafiq syndrome, 614202 (3), Autosomal recessive
MAN2B1	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAN2C1	99.96 %	154580	Congenital disorder of deglycosylation 2, 619775 (3), Autosomal recessive
MANBA	99.81 %	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MAOA	99.83 %	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
MAP1B	100 %	157129	?Deafness, autosomal dominant 83, 619808 (3), Autosomal dominant; Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant
MAP2K1	99.98 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	99.99 %	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MAP4K4	99.57 %	604666	*No OMIM phenotypes*
MAPK1	99.84 %	176948	Noonan syndrome 13, 619087 (3), Autosomal dominant
MAPK8IP3	100 %	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
MAPKAPK5	99.95 %	606723	Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive
MAPRE2	99.96 %	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MARCHF6	99.97 %	613297	Epilepsy, familial adult myoclonic, 3, 613608 (3), Autosomal dominant
MARK2	99.91 %	600526	*No OMIM phenotypes*
MASP1	99.99 %	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MAST1	100 %	612256	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant
MAST3	97.04 %	612258	Developmental and epileptic encephalopathy 108, 620115 (3), Autosomal dominant
MAST4	99.98 %	618002	*No OMIM phenotypes*
MAT1A	99.7 %	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal dominant, Autosomal recessive; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal dominant, Autosomal recessive
MBD5	99.79 %	611472	Intellectual developmental disorder, autosomal dominant 1, 156200 (3), Autosomal dominant
MBOAT7	100 %	606048	Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive
MBTPS2	99.81 %	300294	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MCCC1	99.86 %	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
MCCC2	99.97 %	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
MCM3AP	99.98 %	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
MCOLN1	100 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	100 %	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MDH1	99.96 %	154200	?Developmental and epileptic encephalopathy 88, 618959 (3), Autosomal recessive
MDH2	99.54 %	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
MECP2	99.95 %	300005	Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MECR	99.63 %	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive; Optic atrophy 16, 620629 (3), Autosomal recessive
MED11	99.99 %	612383	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327 (3), Autosomal recessive
MED12	99.94 %	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MED12L	99.95 %	611318	Nizon-Isidor syndrome, 618872 (3), Autosomal dominant
MED13	99.55 %	603808	Intellectual developmental disorder, autosomal dominant 61, 618009 (3), Autosomal dominant
MED13L	99.99 %	608771	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant
MED17	99.82 %	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MED23	99.82 %	605042	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 (3), Autosomal recessive
MED25	99.95 %	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive
MED27	99.99 %	605044	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive
MEF2C	99.57 %	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 (3), Autosomal dominant
MEGF8	99.9 %	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEIS2	99.97 %	601740	Cleft palate, cardiac defects, and impaired intellectual development, 600987 (3), Autosomal dominant
METTL23	100 %	615262	Intellectual developmental disorder, autosomal recessive 44, 615942 (3), Autosomal recessive
METTL5	99.75 %	618628	Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive
MFF	99.97 %	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
MFSD2A	99.78 %	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive
MFSD8	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MGAT2	100 %	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
MICAL1	99.99 %	607129	*No OMIM phenotypes*
MICU1	99.56 %	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MID1	99.9 %	300552	Opitz GBBB syndrome, 300000 (3), X-linked recessive
MINPP1	99.73 %	605391	{Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive
MKKS	100 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKS1	99.92 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MLC1	99.99 %	605908	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive
MLYCD	99.95 %	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
MMAA	99.95 %	607481	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive
MMAB	99.99 %	607568	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive
MMACHC	99.98 %	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	99.76 %	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
MMUT	99.68 %	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MN1	99.98 %	156100	CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant
MOCS1	99.95 %	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MOCS2	99.96 %	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
MOGS	100 %	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MORC2	100 %	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant; Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 (3), Autosomal dominant
MPDU1	99.97 %	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPDZ	99.86 %	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
MPLKIP	99.99 %	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MRAS	99.97 %	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
MRPL3	99.94 %	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
MRPS22	99.87 %	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
MRPS34	100 %	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
MRTFB	99.89 %	609463	*No OMIM phenotypes*
MSL2	100 %	614802	*No OMIM phenotypes*
MSL3	99.83 %	300609	Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant
MSMO1	99.88 %	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
MTFMT	99.98 %	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
MTHFR	99.97 %	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
MTHFS	100 %	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive
MTO1	90.25 %	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
MTOR	99.98 %	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant
MTR	99.95 %	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
MTRFR	99.87 %	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
MTRR	99.98 %	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTSS2	100 %	616951	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086 (3), Autosomal dominant
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYCBP2	99.88 %	610392	*No OMIM phenotypes*
MYCN	100 %	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant; Megalencephaly-polydactyly syndrome, 620748 (3), Autosomal dominant
MYH10	99.91 %	160776	*No OMIM phenotypes*
MYO5A	99.94 %	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MYT1L	99.99 %	613084	Intellectual developmental disorder, autosomal dominant 39, 616521 (3), Autosomal dominant
NAA10	99.99 %	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NAA15	99.71 %	608000	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant
NAA20	99.93 %	610833	Intellectual developmental disorder, autosomal recessive 73, 619717 (3), Autosomal recessive
NACC1	99.99 %	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
NAE1	99.81 %	603385	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210 (3), Autosomal recessive
NAGA	100 %	104170	Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	100 %	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NAGS	99.99 %	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
NALCN	99.97 %	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive
NANS	100 %	605202	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (3), Autosomal recessive
NAPB	100 %	611270	Developmental and epileptic encephalopathy 107, 620033 (3), Autosomal recessive
NARS1	99.94 %	108410	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 (3), Autosomal recessive
NARS2	99.59 %	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive
NAV3	99.08 %	611629	*No OMIM phenotypes*
NAXD	99.99 %	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
NAXE	99.99 %	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
NBEA	99.98 %	604889	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 (3), Autosomal dominant
NBN	99.93 %	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCAPG2	99.97 %	608532	Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive
NCDN	99.99 %	608458	Neurodevelopmental disorder with infantile epileptic spasms, 619373 (3), Autosomal dominant
NCKAP1	99.62 %	604891	*No OMIM phenotypes*
NCOR1	99.88 %	600849	*No OMIM phenotypes*
NDE1	100 %	609449	Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive
NDP	99.98 %	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
NDST1	100 %	600853	Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NDUFA1	99.93 %	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
NDUFA10	99.98 %	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
NDUFA11	98.22 %	612638	Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive
NDUFA12	99.21 %	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFA2	99.95 %	602137	Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
NDUFA6	99.96 %	602138	Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive
NDUFA8	100 %	603359	Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive
NDUFAF1	100 %	606934	Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive
NDUFAF2	99.88 %	609653	Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive
NDUFAF3	100 %	612911	Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive
NDUFAF4	99.95 %	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
NDUFAF5	99.89 %	612360	Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive
NDUFAF6	99.86 %	612392	Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive
NDUFAF8	99.98 %	618461	Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive
NDUFB3	99.6 %	603839	Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive
NDUFB9	100 %	601445	?Mitochondrial complex I deficiency, nuclear type 24, 618245 (3), Autosomal recessive
NDUFS1	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
NDUFS2	99.66 %	602985	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
NDUFS3	100 %	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
NDUFS4	99.99 %	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive
NDUFS6	100 %	603848	Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive
NDUFS7	99.99 %	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
NDUFS8	100 %	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
NDUFV1	99.99 %	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
NDUFV2	99.98 %	600532	Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive
NECAP1	100 %	611623	Developmental and epileptic encephalopathy 21, 615833 (3), Autosomal recessive
NECTIN1	99.99 %	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NEDD4L	99.97 %	606384	Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant
NEMF	99.94 %	608378	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 (3), Autosomal recessive
NEU1	99.98 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
NEUROD2	100 %	601725	Developmental and epileptic encephalopathy 72, 618374 (3), Autosomal dominant
NEUROG1	100 %	601726	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469 (3), Autosomal recessive
NEXMIF	99.99 %	300524	Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant
NF1	99.88 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NFASC	99.94 %	609145	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive
NFE2L2	99.97 %	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
NFIA	97.55 %	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant
NFIB	99.9 %	600728	Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant
NFIX	99.99 %	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant
NFU1	99.48 %	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
NGLY1	99.93 %	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
NHLRC1	100 %	608072	Myoclonic epilepsy of Lafora 2, 620681 (3), Autosomal recessive
NHLRC2	99.83 %	618277	FINCA syndrome, 618278 (3), Autosomal recessive
NHS	99.96 %	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
NIPBL	99.34 %	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKAP	99.1 %	300766	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive
NKX2-1	100 %	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NLGN3	99.98 %	300336	{Autism susceptibility, X-linked 1}, 300425 (3), X-linked
NLGN4X	99.98 %	300427	Intellectual developmental disorder, X-linked, 300495 (3), X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), X-linked
NLRP3	100 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NONO	99.94 %	300084	Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked
NOVA2	99.95 %	601991	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	99.05 %	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPRL2	100 %	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
NPRL3	99.99 %	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant
NPTN	99.93 %	612820	*No OMIM phenotypes*
NR2F1	99.99 %	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
NR2F2	100 %	107773	46XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NR4A2	99.97 %	601828	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NRAS	99.66 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NRCAM	99.82 %	601581	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive
NRROS	100 %	615322	Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive
NRXN1	99.98 %	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
NRXN2	99.98 %	600566	*No OMIM phenotypes*
NSD1	99.98 %	606681	Sotos syndrome, 117550 (3), Autosomal dominant
NSD2	99.87 %	602952	Rauch-Steindl syndrome, 619695 (3), Autosomal dominant
NSDHL	99.87 %	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
NSF	52.85 %	601633	Developmental and epileptic encephalopathy 96, 619340 (3), Autosomal dominant
NSRP1	99.96 %	616173	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive
NSUN2	99.96 %	610916	Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive
NSUN6	99.95 %	617199	Intellectual developmental disorder, autosomal recessive 82, 620779 (3), Autosomal recessive
NT5C2	99.96 %	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
NTNG2	99.98 %	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
NTRK1	99.86 %	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
NTRK2	99.9 %	600456	Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant
NUBPL	99.62 %	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
NUDT2	100 %	602852	Intellectual developmental disorder with or without peripheral neuropathy, 619844 (3), Autosomal recessive
NUP107	97.46 %	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
NUP188	99.88 %	615587	Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive
NUP214	99.98 %	114350	Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive
NUP62	100 %	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
NUP85	99.98 %	170285	Nephrotic syndrome, type 17, 618176 (3), Autosomal recessive
NUS1	99.9 %	610463	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
OAT	90.17 %	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OCLN	82.91 %	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OCRL	99.89 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
ODC1	99.99 %	165640	Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant
OFD1	99.68 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
OGDH	99.93 %	613022	Oxoglutarate dehydrogenase deficiency, 203740 (3), Autosomal recessive
OGDHL	99.95 %	617513	Yoon-Bellen neurodevelopmental syndrome, 619701 (3), Autosomal recessive
OGT	99.79 %	300255	Intellectual developmental disorder, X-linked 106, 300997 (3), X-linked recessive
OPHN1	99.92 %	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive
ORC1	99.7 %	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
OSGEP	100 %	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
OTC	99.42 %	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
OTUD5	99.93 %	300713	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive
OTUD6B	99.92 %	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
OTUD7A	99.04 %	612024	Neurodevelopmental disorder with hypotonia and seizures, 620790 (3), Autosomal recessive
OTX2	100 %	600037	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
OXR1	99.96 %	605609	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive
P4HTM	100 %	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive
PABPC1	99.84 %	604679	*No OMIM phenotypes*
PACS1	99.96 %	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PACS2	99.99 %	610423	Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant
PAFAH1B1	99.96 %	601545	Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant
PAH	99.96 %	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PAK1	99.99 %	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant
PAK2	99.83 %	605022	?Knobloch syndrome 2, 618458 (3), Autosomal dominant
PAK3	92.53 %	300142	Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive
PALS1	99.82 %	606958	*No OMIM phenotypes*
PAN2	99.95 %	617447	*No OMIM phenotypes*
PANK2	99.99 %	606157	Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PARN	99.75 %	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant
PARP6	99.97 %	619439	*No OMIM phenotypes*
PARS2	99.99 %	612036	Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive
PAX1	100 %	167411	Otofaciocervical syndrome 2 with T-cell deficiency, 615560 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PAX5	99.82 %	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PAX6	99.95 %	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; Microphthalmia/coloboma 12, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
PAX8	99.99 %	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PBX1	99.88 %	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
PC	99.99 %	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCCA	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH12	100 %	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCDH19	99.98 %	300460	Developmental and epileptic encephalopathy 9, 300088 (3), X-linked
PCDHGC4	100 %	606305	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 (3), Autosomal recessive
PCGF2	99.78 %	600346	Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant
PCLO	99.41 %	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
PCNT	99.97 %	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PCYT2	100 %	602679	Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive
PDCD10	99.94 %	609118	Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant
PDE10A	87.37 %	610652	Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive
PDE2A	99.95 %	602658	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive
PDE4D	99.89 %	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
PDE6D	99.94 %	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive
PDGFRB	99.99 %	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant
PDHA1	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHB	99.94 %	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
PDHX	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PDP1	100 %	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
PDSS1	95.7 %	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	99.87 %	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
PDZD8	99.99 %	614235	Intellectual developmental disorder with autism and dysmorphic facies, 620021 (3), Autosomal recessive
PEPD	99.98 %	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PET100	99.98 %	614770	Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PEX1	98.8 %	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
PEX10	100 %	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	99.62 %	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
PEX12	100 %	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
PEX13	99.36 %	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	100 %	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	99.94 %	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
PEX19	99.25 %	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX2	100 %	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX26	100 %	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive
PEX3	99.85 %	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
PEX5	99.89 %	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX6	99.99 %	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
PEX7	99.72 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PGAP1	99.56 %	611655	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive
PGAP2	99.99 %	615187	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207 (3), Autosomal recessive
PGAP3	99.97 %	611801	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716 (3), Autosomal recessive
PGK1	99.93 %	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM2L1	99.82 %	611610	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191 (3), Autosomal recessive
PGM3	99.94 %	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHACTR1	100 %	608723	Developmental and epileptic encephalopathy 70, 618298 (3), Autosomal dominant
PHF21A	99.91 %	608325	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant
PHF5A	100 %	617846	*No OMIM phenotypes*
PHF6	99.16 %	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
PHF8	99.87 %	300560	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive
PHGDH	99.79 %	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHIP	99.63 %	612870	Chung-Jansen syndrome, 617991 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PI4K2A	99.85 %	609763	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732 (3), Autosomal recessive
PI4KA	99.76 %	600286	Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PIBF1	99.9 %	607532	Joubert syndrome 33, 617767 (3), Autosomal recessive
PIDD1	100 %	605247	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 (3), Autosomal recessive
PIGA	99.81 %	311770	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3), X-linked recessive
PIGB	99.92 %	604122	Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive
PIGC	100 %	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
PIGF	98.59 %	600153	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 (3), Autosomal recessive
PIGG	99.99 %	616918	[Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive
PIGH	100 %	600154	Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive
PIGK	92.56 %	605087	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive
PIGL	99.98 %	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGM	99.97 %	610273	Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive
PIGN	99.91 %	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
PIGO	99.99 %	614730	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 (3), Autosomal recessive
PIGP	99.87 %	605938	Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive
PIGQ	99.99 %	605754	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 (3), Autosomal recessive
PIGS	100 %	610271	Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive
PIGT	99.95 %	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
PIGU	99.98 %	608528	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive
PIGV	100 %	610274	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 (3), Autosomal recessive
PIGW	99.87 %	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
PIGY	99.99 %	610662	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809 (3), Autosomal recessive
PIK3C2B	99.79 %	602838	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PIK3CA	99.74 %	171834	Hemifacial myohyperplasia, somatic, 606773 (3); CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3)
PIK3R2	99.95 %	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
PIP5K1C	99.94 %	606102	Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive
PISD	100 %	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive
PITRM1	99.89 %	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
PLA2G6	99.98 %	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
PLAA	99.79 %	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
PLCB1	99.98 %	607120	Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive
PLCH1	99.99 %	612835	Holoprosencephaly 14, 619895 (3), Autosomal recessive
PLEKHG2	99.99 %	611893	Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive
PLK1	99.97 %	602098	*No OMIM phenotypes*
PLK4	99.89 %	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PLP1	99.98 %	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PLPBP	99.99 %	604436	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290 (3), Autosomal recessive
PLXNA1	100 %	601055	Dworschak-Punetha neurodevelopmental syndrome, 619955 (3), Autosomal recessive
PLXNB2	100 %	604293	*No OMIM phenotypes*
PLXND1	99.98 %	604282	Congenital heart defects, multiple types, 9, 620294 (3), Autosomal recessive
PMM2	99.93 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMPCA	99.99 %	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
PMPCB	99.94 %	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
PNKP	100 %	605610	?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNP	100 %	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
PNPLA6	99.99 %	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
PNPLA8	99.92 %	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
PNPO	99.9 %	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PNPT1	99.56 %	610316	Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
POC1A	99.98 %	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POC1B	100 %	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
POGZ	99.49 %	614787	White-Sutton syndrome, 616364 (3), Autosomal dominant
POLA1	99.57 %	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
POLG	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLG2	99.51 %	604983	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive
POLR1A	99.93 %	616404	Leukodystrophy, hypomyelinating, 27, 620675 (3), Autosomal recessive; Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant
POLR1C	100 %	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR2A	99.95 %	180660	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant
POLR3A	99.97 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	99.94 %	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
POLRMT	99.99 %	601778	Combined oxidative phosphorylation deficiency 55, 619743 (3), Autosomal dominant, Autosomal recessive
POMGNT1	99.69 %	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
POMGNT2	100 %	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
POMK	100 %	615247	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
POMT1	99.96 %	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive
POMT2	99.98 %	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive
PORCN	99.93 %	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
POU1F1	99.98 %	173110	Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal dominant, Autosomal recessive
POU3F2	99.81 %	600494	*No OMIM phenotypes*
POU3F3	99.96 %	602480	Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant
PPFIA2	99.43 %	603143	*No OMIM phenotypes*
PPFIA3	99.97 %	603144	*No OMIM phenotypes*
PPFIBP1	99.11 %	603141	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive
PPIL1	100 %	601301	Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive
PPM1D	99.81 %	605100	Breast cancer, somatic, 114480 (3); Jansen-de Vries syndrome, 617450 (3), Autosomal dominant
PPOX	99.85 %	600923	Variegate porphyria, childhood-onset, 620483 (3), Autosomal recessive; Variegate porphyria, 176200 (3), Autosomal dominant
PPP1CB	99.89 %	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PPP1R12A	99.08 %	602021	Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant
PPP1R15B	99.92 %	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
PPP1R21	99.92 %	618159	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive
PPP1R3F	99.83 %	301104	*No OMIM phenotypes*
PPP2CA	99.99 %	176915	Houge-Janssens syndrome 3, 618354 (3), Autosomal dominant
PPP2R1A	100 %	605983	Houge-Janssens syndrome 2, 616362 (3), Autosomal dominant
PPP2R5C	99.96 %	601645	*No OMIM phenotypes*
PPP2R5D	99.99 %	601646	Houge-Janssens syndrome 1, 616355 (3), Autosomal dominant
PPP3CA	99.84 %	114105	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant
PPT1	97.48 %	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PQBP1	99.99 %	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRDM13	99.99 %	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
PRDM15	99.99 %	617692	*No OMIM phenotypes*
PRDM8	99.99 %	616639	?Epilepsy, progressive myoclonic, 10, 616640 (3), Autosomal recessive
PREPL	99.63 %	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PRF1	100 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRICKLE1	99.87 %	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRICKLE2	99.98 %	608501	*No OMIM phenotypes*
PRKACB	87.33 %	176892	Cardioacrofacial dysplasia 2, 619143 (3), Somatic mosaicism, Autosomal dominant
PRKAR1A	100 %	188830	Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant
PRKAR1B	100 %	176911	Marbach-Schaaf neurodevelopmental syndrome, 619680 (3), Autosomal dominant
PRKD1	99.96 %	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PRMT7	99.95 %	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PRODH	4.29 %	606810	{Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive
PRORP	99.8 %	609947	Combined oxidative phosphorylation deficiency 54, 619737 (3), Autosomal recessive
PRPF19	99.99 %	608330	*No OMIM phenotypes*
PRPF8	99.98 %	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant
PRPS1	99.95 %	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
PRR12	100 %	616633	Neuroocular syndrome, 619539 (3), Autosomal dominant
PRRT2	99.97 %	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant
PRSS12	99.98 %	606709	Intellectual developmental disorder, autosomal recessive 1, 249500 (3), Autosomal recessive
PRUNE1	99.85 %	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
PSAP	99.94 %	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
PSAT1	99.98 %	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSMB1	99.99 %	602017	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038 (3), Autosomal recessive
PSMC3	99.93 %	186852	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354 (3), Autosomal recessive
PSMC5	100 %	601681	*No OMIM phenotypes*
PSMD11	99.95 %	604449	*No OMIM phenotypes*
PSMD12	99.95 %	604450	Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant
PSPH	99.09 %	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PTBP1	100 %	600693	*No OMIM phenotypes*
PTCD3	99.77 %	614918	Combined oxidative phosphorylation deficiency 51, 619057 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PTCH1	99.99 %	601309	Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTCHD1	99.97 %	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
PTDSS1	99.95 %	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTEN	99.89 %	601728	{Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
PTF1A	100 %	607194	Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive
PTPA	100 %	600756	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482 (3), Autosomal recessive
PTPN11	99.98 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTPN23	100 %	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
PTPN4	98.2 %	176878	*No OMIM phenotypes*
PTRH2	99.99 %	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PTRHD1	100 %	617342	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747 (3), Autosomal recessive
PTS	99.93 %	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
PUF60	100 %	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PUM1	98.14 %	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant; Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719 (3), Autosomal dominant
PURA	100 %	600473	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 (3), Autosomal dominant
PUS1	100 %	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PUS3	100 %	616283	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive
PUS7	99.89 %	616261	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive
PYCR1	99.99 %	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
PYCR2	99.95 %	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
QARS1	100 %	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
QDPR	99.92 %	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
QRICH1	100 %	617387	Ververi-Brady syndrome, 617982 (3), Autosomal dominant
RAB11A	100 %	605570	*No OMIM phenotypes*
RAB11B	100 %	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
RAB18	99.76 %	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB23	99.97 %	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAB27A	99.94 %	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RAB39B	99.99 %	300774	Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive
RAB3GAP1	99.73 %	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	99.69 %	609275	Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RAB5C	99.99 %	604037	*No OMIM phenotypes*
RABGAP1	99.99 %	615882	*No OMIM phenotypes*
RAC1	99.75 %	602048	Intellectual developmental disorder, autosomal dominant 48, 617751 (3), Autosomal dominant
RAC3	100 %	602050	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant
RAD21	99.91 %	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive
RAF1	99.97 %	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant
RAI1	99.22 %	607642	Smith-Magenis syndrome, 182290 (3), Isolated cases, Autosomal dominant
RALA	99.69 %	179550	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant
RALGAPA1	99.73 %	608884	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive
RAP1B	98.57 %	179530	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654 (3), Autosomal dominant
RAP1GDS1	99.8 %	179502	Alfadhel syndrome, 620655 (3), Autosomal recessive
RAPGEF2	99.84 %	609530	?Epilepsy, familial adult myoclonic, 7, 618075 (3), Autosomal dominant
RARB	99.99 %	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal dominant, Autosomal recessive
RARS1	99.76 %	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
RARS2	99.88 %	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RBBP5	99.89 %	600697	*No OMIM phenotypes*
RBBP8	99.91 %	604124	Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3)
RBL2	99.44 %	180203	Brunet-Wagner neurodevelopmental syndrome, 619690 (3), Autosomal recessive
RBM10	99.98 %	300080	TARP syndrome, 311900 (3), X-linked recessive
RBM28	99.99 %	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBPJ	99.96 %	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RBSN	100 %	609511	*No OMIM phenotypes*
RCBTB1	99.99 %	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RELN	99.98 %	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
RERE	99.94 %	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
RFT1	99.79 %	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RFX3	99.95 %	601337	*No OMIM phenotypes*
RFX4	99.95 %	603958	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RFX7	99.99 %	612660	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330 (3), Autosomal dominant
RHEB	99.97 %	601293	*No OMIM phenotypes*
RHOBTB2	100 %	607352	Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant
RIMS2	99.97 %	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
RIT1	99.78 %	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RLIM	99.93 %	300379	Tonne-Kalscheuer syndrome, 300978 (3), X-linked
RMND1	99.92 %	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
RNASEH2A	99.95 %	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	99.94 %	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	99.99 %	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNASET2	99.99 %	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
RNF113A	99.99 %	300951	Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked
RNF125	99.98 %	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
RNF13	99.81 %	609247	Developmental and epileptic encephalopathy 73, 618379 (3), Autosomal dominant
RNF220	99.68 %	616136	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive
RNPC3	88.9 %	618016	Pituitary hormone deficiency, combined or isolated, 7, 618160 (3), Autosomal recessive
RNU4ATAC	99.95 %	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
RNU7-1	33.9 %	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
ROBO1	99.83 %	602430	Pituitary hormone deficiency, combined or isolated, 8, 620303 (3), Autosomal dominant; Neurooculorenal syndrome, 620305 (3), Autosomal recessive; ?Nystagmus 8, congenital, autosomal recessive, 257400 (3), Autosomal recessive
ROGDI	99.98 %	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
RORA	99.97 %	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant
RORB	99.86 %	601972	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 (3), Autosomal dominant
RPGRIP1L	96.35 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
RPH3A	99.99 %	612159	*No OMIM phenotypes*
RPIA	99.84 %	180430	Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive
RPL10	84.91 %	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive
RPS19	100 %	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS6KA3	98.93 %	300075	Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant
RRAS	99.98 %	165090	*No OMIM phenotypes*
RRAS2	99.94 %	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RRM2B	99.97 %	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
RRP7A	97.99 %	619449	?Microcephaly 28, primary, autosomal recessive, 619453 (3), Autosomal recessive
RSPRY1	99.73 %	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
RSRC1	99.95 %	613352	Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive
RTEL1	100 %	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant
RTN4IP1	99.98 %	610502	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive
RTTN	99.93 %	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
RUBCN	100 %	613516	Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
RUSC2	99.99 %	611053	Intellectual developmental disorder, autosomal recessive 61, 617773 (3), Autosomal recessive
RXYLT1	99.48 %	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
RYR3	99.98 %	180903	Congenital myopathy 20, 620310 (3), Autosomal recessive
SALL1	100 %	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SAMD12	100 %	618073	Epilepsy, familial adult myoclonic, 1, 601068 (3), Autosomal dominant
SAMD9	99.93 %	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SAMHD1	99.98 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SARS1	98.53 %	607529	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive
SARS2	99.99 %	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
SART3	99.99 %	611684	*No OMIM phenotypes*
SASS6	95.06 %	609321	Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive
SATB1	100 %	602075	den Hoed-de Boer-Voisin syndrome, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant
SATB2	99.96 %	608148	Glass syndrome, 612313 (3), Autosomal dominant
SBDS	99.93 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SBF1	99.99 %	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive
SC5D	99.97 %	602286	Lathosterolosis, 607330 (3), Autosomal recessive
SCAF4	99.91 %	616023	Fliedner-Zweier syndrome, 620511 (3), Autosomal dominant
SCAMP5	99.99 %	613766	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SCAPER	99.75 %	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
SCARB2	99.99 %	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCN1A	99.94 %	182389	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant
SCN1B	99.98 %	600235	Generalized epilepsy with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Developmental and epileptic encephalopathy 52, 617350 (3), Autosomal recessive; Cardiac conduction defect, nonspecific, 612838 (3); Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3)
SCN2A	99.86 %	182390	Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant
SCN3A	99.76 %	182391	Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant
SCN8A	99.77 %	600702	?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant
SCO1	99.98 %	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
SCO2	100 %	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
SCYL1	100 %	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SDCCAG8	100 %	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SDHA	99.98 %	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Pheochromocytoma/paraganglioma syndrome 5, 614165 (3), Autosomal dominant
SDHAF1	99.99 %	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
SEC31A	99.86 %	610257	?Halperin-Birk syndrome, 618651 (3), Autosomal recessive
SEMA6B	99.98 %	608873	Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant
SEPHS1	100 %	600902	*No OMIM phenotypes*
SEPSECS	99.78 %	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SERAC1	99.9 %	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SERPINI1	99.98 %	602445	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant
SET	78.28 %	600960	Intellectual developmental disorder, autosomal dominant 58, 618106 (3), Autosomal dominant
SETBP1	100 %	611060	Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant
SETD1A	99.99 %	611052	Epilepsy, early-onset, 2, with or without developmental delay, 618832 (3), Autosomal dominant; Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SETD1B	99.99 %	611055	Intellectual developmental disorder with seizures and language delay, 619000 (3), Autosomal dominant
SETD2	99.91 %	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 70, 620157 (3), Autosomal dominant; Rabin-Pappas syndrome, 620155 (3), Autosomal dominant
SETD5	99.99 %	615743	Intellectual developmental disorder, autosomal dominant 23, 615761 (3), Autosomal dominant
SF1	99.88 %	601516	*No OMIM phenotypes*
SFXN4	99.97 %	615564	Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive
SGPL1	99.95 %	603729	RENI syndrome, 617575 (3), Autosomal recessive
SGSH	100 %	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SGSM3	100 %	610440	*No OMIM phenotypes*
SHANK1	99.99 %	604999	*No OMIM phenotypes*
SHANK2	99.97 %	603290	{Autism susceptibility 17}, 613436 (3)
SHANK3	98.45 %	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant
SHH	100 %	600725	Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant
SHMT2	99.92 %	138450	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive
SHOC2	99.96 %	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SHQ1	99.81 %	613663	Neurodevelopmental disorder with dystonia and seizures, 619922 (3), Autosomal recessive; ?Dystonia 35, childhood-onset, 619921 (3), Autosomal recessive
SIAH1	100 %	602212	Buratti-Harel syndrome, 619314 (3), Autosomal dominant
SIK1	3.83 %	605705	Developmental and epileptic encephalopathy 30, 616341 (3), Autosomal dominant
SIL1	99.95 %	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIN3A	99.97 %	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
SIN3B	99.98 %	607777	*No OMIM phenotypes*
SIX3	100 %	603714	Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant
SKI	99.98 %	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SLC12A2	99.55 %	600840	Kilquist syndrome, 619080 (3), Autosomal recessive; Delpire-McNeill syndrome, 619083 (3), Autosomal dominant; Deafness, autosomal dominant 78, 619081 (3), Autosomal dominant
SLC12A5	99.99 %	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive
SLC12A6	99.98 %	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2II, 620068 (3), Autosomal dominant
SLC13A3	99.98 %	606411	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive
SLC13A5	99.99 %	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive
SLC16A1	99.26 %	600682	Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC16A2	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC17A5	99.71 %	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC19A3	99.95 %	606152	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 (3), Autosomal recessive
SLC1A1	99.98 %	133550	Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3)
SLC1A2	99.95 %	600300	Developmental and epileptic encephalopathy 41, 617105 (3), Autosomal dominant
SLC1A4	99.97 %	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
SLC25A1	99.93 %	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive
SLC25A12	99.72 %	603667	Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive
SLC25A15	100 %	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A19	99.99 %	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
SLC25A20	100 %	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC25A22	100 %	609302	Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive
SLC25A26	99.76 %	611037	Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive
SLC25A42	99.99 %	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
SLC2A1	99.93 %	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant
SLC30A9	99.75 %	604604	Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive
SLC32A1	100 %	616440	Generalized epilepsy with febrile seizures plus, type 12, 620755 (3), Autosomal dominant; Developmental and epileptic encephalopathy 114, 620774 (3), Autosomal dominant
SLC33A1	99.67 %	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Huppke-Brendel syndrome, 614482 (3), Autosomal recessive
SLC35A1	99.81 %	605634	Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive
SLC35A2	99.97 %	314375	Congenital disorder of glycosylation, type IIm, 300896 (3), Somatic mosaicism, X-linked dominant
SLC35A3	94.67 %	605632	Arthrogryposis, impaired intellectual development, and seizures, 615553 (3), Autosomal recessive
SLC35B2	100 %	610788	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 (3), Autosomal recessive
SLC35C1	100 %	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC38A3	100 %	604437	Developmental and epileptic encephalopathy 102, 619881 (3), Autosomal recessive
SLC39A14	92.87 %	608736	?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive
SLC39A8	99.95 %	608732	Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC45A1	99.99 %	605763	Intellectual developmental disorder with neuropsychiatric features, 617532 (3), Autosomal recessive
SLC46A1	100 %	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC4A10	99.69 %	605556	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746 (3), Autosomal recessive
SLC4A4	99.97 %	603345	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 (3), Autosomal recessive
SLC5A6	100 %	604024	Sodium-dependent multivitamin transporter deficiency, 618973 (3), Autosomal recessive; Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 (3), Autosomal recessive
SLC6A1	99.96 %	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
SLC6A17	99.79 %	610299	Intellectual developmental disorder, autosomal recessive 48, 616269 (3), Autosomal recessive
SLC6A19	99.99 %	608893	Hartnup disorder, 234500 (3), Autosomal recessive
SLC6A3	99.96 %	126455	Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3)
SLC6A8	99.99 %	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SLC6A9	99.93 %	601019	Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive
SLC7A6OS	99.96 %	619192	Epilepsy, progressive myoclonic, 12, 619191 (3), Autosomal recessive
SLC7A7	99.99 %	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC9A6	99.42 %	300231	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked
SLC9A7	99.91 %	300368	Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive
SLF2	99.89 %	610348	Atelis syndrome 1, 620184 (3), Autosomal recessive
SLITRK2	100 %	300561	Intellectual developmental disorder, X-linked 111, 301107 (3), X-linked
SLX4	100 %	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMAD4	99.97 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMARCA1	97.35 %	300012	*No OMIM phenotypes*
SMARCA2	99.95 %	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant
SMARCA4	99.99 %	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant
SMARCA5	99.81 %	603375	*No OMIM phenotypes*
SMARCB1	99.99 %	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant
SMARCC2	99.73 %	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
SMARCD1	99.81 %	601735	Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant
SMARCE1	99.87 %	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SMC1A	99.98 %	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant
SMC3	99.91 %	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SMC5	99.47 %	609386	Atelis syndrome 2, 620185 (3), Autosomal recessive
SMG8	99.92 %	613175	Alzahrani-Kuwahara syndrome, 619268 (3), Autosomal recessive
SMG9	99.99 %	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive; Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995 (3), Autosomal recessive
SMOC1	100 %	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMPD1	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SMPD4	99.9 %	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive
SMS	98.56 %	300105	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive
SNAP25	99.89 %	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SNAP29	99.85 %	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNAPC4	100 %	602777	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 (3), Autosomal recessive
SNF8	99.78 %	610904	Developmental and epileptic encephalopathy 115, 620783 (3), Autosomal recessive; Neurodevelopmental disorder plus optic atrophy, 620784 (3), Autosomal recessive
SNIP1	99.96 %	608241	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive
SNORD118	100 %	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
SNRPN	99.97 %	182279	*No OMIM phenotypes*
SNX14	99.73 %	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
SNX27	99.59 %	611541	*No OMIM phenotypes*
SOBP	99.99 %	613667	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
SON	99.95 %	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
SOS1	99.68 %	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	99.39 %	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX10	100 %	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant
SOX11	100 %	600898	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 (3), Autosomal dominant
SOX2	100 %	184429	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant
SOX3	100 %	313430	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX4	99.36 %	184430	Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant
SOX5	99.96 %	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
SOX6	99.89 %	607257	Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant
SP9	100 %		*No OMIM phenotypes*
SPART	99.98 %	607111	Troyer syndrome, 275900 (3), Autosomal recessive
SPAST	99.77 %	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SPATA5	99.82 %	613940	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive
SPATA5L1	99.91 %	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
SPECC1L	99.98 %	614140	Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant
SPEN	99.98 %	613484	Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant
SPG11	99.89 %	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPOP	99.85 %	602650	Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant
SPOUT1	100 %	617614	*No OMIM phenotypes*
SPR	99.99 %	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SPRED1	99.99 %	609291	Legius syndrome, 611431 (3), Autosomal dominant
SPRED2	99.99 %	609292	Noonan syndrome 14, 619745 (3), Autosomal recessive
SPTAN1	99.96 %	182810	Developmental delay with or without epilepsy, 620540 (3), Autosomal dominant; Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant; Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528 (3), Autosomal dominant
SPTBN1	99.98 %	182790	Developmental delay, impaired speech, and behavioral abnormalities, 619475 (3), Autosomal dominant
SPTBN2	99.98 %	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SPTBN4	99.91 %	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
SRCAP	99.99 %	611421	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 (3), Autosomal dominant; Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRD5A3	99.94 %	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
SRP54	99.83 %	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
SRPK3	99.98 %	301002	*No OMIM phenotypes*
SRRM2	99.98 %	606032	Intellectual developmental disorder, autosomal dominant 72, 620439 (3), Autosomal dominant
SRSF1	99.99 %	600812	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489 (3), Autosomal dominant
SSR4	99.99 %	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
ST3GAL3	99.98 %	606494	Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive
ST3GAL5	99.96 %	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAG1	99.88 %	604358	Intellectual developmental disorder, autosomal dominant 47, 617635 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
STAG2	99.08 %	300826	Holoprosencephaly 13, X-linked, 301043 (3), X-linked recessive, X-linked dominant; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked
STAMBP	99.95 %	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
STARD7	99.6 %	616712	Epilepsy, familial adult myoclonic, 2, 607876 (3), Autosomal dominant
STEEP1	99.89 %	301012	?Intellectual developmental disorder, X-linked 107, 301013 (3), X-linked
STIL	99.11 %	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STRA6	99.95 %	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
STRADA	99.98 %	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
STT3A	99.99 %	601134	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive
STT3B	99.96 %	608605	Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
STX1A	99.98 %	186590	*No OMIM phenotypes*
STX1B	99.97 %	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
STXBP1	99.99 %	602926	Developmental and epileptic encephalopathy 4, 612164 (3), Autosomal dominant, Autosomal recessive
SUCLA2	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	99.64 %	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUFU	100 %	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal dominant, Autosomal recessive
SUMF1	99.95 %	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUOX	100 %	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SUPT16H	99.97 %	605012	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 (3), Autosomal dominant
SURF1	100 %	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
SUZ12	98.58 %	606245	Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant
SV2A	99.65 %	185860	Developmental and epileptic encephalopathy 113, 620772 (3), Autosomal recessive
SVBP	99.04 %	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
SYN1	99.98 %	313440	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491 (3), X-linked; Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked
SYNCRIP	99.93 %	616686	*No OMIM phenotypes*
SYNE1	99.95 %	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYNGAP1	99.96 %	603384	Intellectual developmental disorder, autosomal dominant 5, 612621 (3), Autosomal dominant
SYNJ1	99.91 %	604297	Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive
SYP	99.99 %	313475	Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SYT1	99.7 %	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
SYT14	99.94 %	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
SZT2	99.88 %	615463	Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive
TAB2	99.75 %	605101	Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant
TACO1	100 %	612958	Mitochondrial complex IV deficiency, nuclear type 8, 619052 (3), Autosomal recessive
TAF1	99.84 %	313650	Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive
TAF13	95.71 %	600774	Intellectual developmental disorder, autosomal recessive 60, 617432 (3), Autosomal recessive
TAF1C	100 %	604905	*No OMIM phenotypes*
TAF2	99.72 %	604912	Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive
TAF4	99.39 %	601796	Intellectual developmental disorder, autosomal dominant 73, 620450 (3), Autosomal dominant
TAF6	99.96 %	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
TAF8	99.91 %	609514	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 (3), Autosomal recessive
TANC2	99.85 %	615047	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant
TANGO2	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TAOK1	99.77 %	610266	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 (3), Autosomal dominant
TARS2	99.77 %	612805	Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive
TASP1	99.86 %	608270	Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive
TAT	99.99 %	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TBC1D20	100 %	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D23	98.7 %	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
TBC1D24	100 %	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
TBC1D2B	99.94 %	619152	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive
TBC1D32	99.75 %	615867	*No OMIM phenotypes*
TBC1D7	99.9 %	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
TBCD	100 %	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
TBCE	99.91 %	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
TBCK	99.75 %	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
TBL1XR1	99.92 %	608628	Intellectual developmental disorder, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TBP	99.98 %	600075	Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
TBR1	99.99 %	604616	Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant
TBX1	99.95 %	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TCEAL1	100 %	300237	Hijazi-Reis syndrome, 301094 (3), X-linked dominant
TCF20	100 %	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant
TCF4	98.56 %	602272	Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant
TCF7L2	99.69 %	602228	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
TCN2	100 %	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TCTN1	99.92 %	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	99.99 %	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	99.92 %	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TDP2	99.97 %	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
TECPR2	99.96 %	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
TECR	99.99 %	610057	Intellectual developmental disorder, autosomal recessive 14, 614020 (3), Autosomal recessive
TEFM	99.88 %	616422	Combined oxidative phosphorylation deficiency 58, 620451 (3), Autosomal recessive
TELO2	99.99 %	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
TENM3	99.99 %	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
TET3	99.95 %	613555	Beck-Fahrner syndrome, 618798 (3), Autosomal dominant, Autosomal recessive
TFAP2A	100 %	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TFE3	99.87 %	314310	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 (3), X-linked; Renal cell carcinoma, papillary, 1, 300854 (3)
TGDS	99.86 %	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
TGIF1	100 %	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
TH	99.99 %	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THG1L	99.99 %	618802	Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive
THOC2	99.11 %	300395	Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive
THOC6	99.94 %	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
THRA	100 %	190120	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant
THRB	99.82 %	190160	Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
THUMPD1	99.79 %	616662	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989 (3), Autosomal recessive
TIAM1	99.99 %	600687	Neurodevelopmental disorder with language delay and seizures, 619908 (3), Autosomal recessive
TIMM50	99.99 %	607381	3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TIMM8A	100 %	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TINF2	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TKT	97.39 %	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
TLK2	98.83 %	608439	Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant
TMCO1	99.48 %	614123	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive
TMEM106B	99.92 %	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
TMEM107	100 %	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM138	100 %	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM147	100 %	613585	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075 (3), Autosomal recessive
TMEM161B	89.66 %		*No OMIM phenotypes*
TMEM163	100 %	618978	Leukodystrophy, hypomyelinating, 25, 620243 (3), Autosomal dominant
TMEM165	99.97 %	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
TMEM216	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM218	99.94 %	619285	Joubert syndrome 39, 619562 (3), Autosomal recessive
TMEM222	99.93 %	619469	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 (3), Autosomal recessive
TMEM231	88.88 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	99.3 %	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM240	99.99 %	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TMEM63A	99.91 %	618685	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant
TMEM63B	99.95 %	619952	*No OMIM phenotypes*
TMEM63C	99.99 %	619953	Spastic paraplegia 87, autosomal recessive, 619966 (3), Autosomal recessive
TMEM67	99.69 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TMEM70	99.99 %	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMEM94	99.96 %	618163	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive
TMLHE	77.23 %	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TMTC3	97.8 %	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
TMX2	99.99 %	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
TNIK	99.93 %	610005	Intellectual developmental disorder, autosomal recessive 54, 617028 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TNPO2	99.99 %	603002	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556 (3), Autosomal dominant
TNR	99.8 %	601995	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 (3), Autosomal recessive
TNRC6A	99.48 %	610739	?Epilepsy, familial adult myoclonic, 6, 618074 (3), Autosomal dominant
TNRC6B	99.99 %	610740	Global developmental delay with speech and behavioral abnormalities, 619243 (3), Autosomal dominant
TOE1	99.96 %	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
TOGARAM1	99.93 %	617618	Joubert syndrome 37, 619185 (3), Autosomal recessive
TOP3A	99.91 %	601243	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive
TOR1A	100 %	605204	{Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant
TP53RK	100 %	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
TP73	100 %	601990	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive
TPI1	99.95 %	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPK1	99.96 %	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TPO	100 %	606765	Thyroid dyshormonogenesis 2A, 274500 (3), Autosomal recessive
TPP1	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TPRKB	81.09 %	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
TRA2B	99.98 %	602719	*No OMIM phenotypes*
TRAF7	99.97 %	606692	Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant
TRAIP	99.97 %	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive
TRAK1	99.98 %	608112	Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive
TRAPPC10	84.42 %	602103	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027 (3), Autosomal recessive
TRAPPC11	99.93 %	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
TRAPPC12	99.96 %	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
TRAPPC2L	100 %	610970	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive
TRAPPC4	100 %	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive
TRAPPC6B	99.74 %	610397	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive
TRAPPC9	99.98 %	611966	Intellectual developmental disorder, autosomal recessive 13, 613192 (3), Autosomal recessive
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TRIM32	100 %	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRIM8	99.82 %	606125	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 (3), Autosomal dominant
TRIO	99.98 %	601893	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 (3), Autosomal dominant
TRIP12	99.62 %	604506	Intellectual developmental disorder, autosomal dominant 49, 617752 (3), Autosomal dominant
TRIP4	99.97 %	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive
TRIT1	99.25 %	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
TRMT1	100 %	611669	Intellectual developmental disorder, autosomal recessive 68, 618302 (3), Autosomal recessive
TRMT10A	99.9 %	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
TRMT10C	99.55 %	615423	Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive
TRMT5	99.99 %	611023	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539 (3), Autosomal recessive
TRNT1	99.97 %	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
TRPM3	99.89 %	608961	?Cataract 50 with or without glaucoma, 620253 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224 (3), Autosomal dominant
TRPM6	99.93 %	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
TRRAP	99.79 %	603015	?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant
TSC1	99.99 %	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	99.98 %	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TSEN15	99.57 %	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
TSEN2	99.98 %	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
TSEN34	100 %	608754	?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive
TSEN54	100 %	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TSFM	100 %	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
TSHB	99.95 %	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
TSPAN7	99.97 %	300096	Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive
TSPOAP1	99.95 %	610764	Dystonia 22, juvenile-onset, 620453 (3), Autosomal recessive; ?Dystonia 22, adult-onset, 620456 (3), Autosomal recessive
TTC19	99.99 %	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTC21B	99.5 %	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
TTC37	99.82 %	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TTC5	100 %	619014	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive
TTC8	99.67 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTI1	100 %	614425	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445 (3), Autosomal recessive
TTI2	99.94 %	614426	Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive
TUBA1A	99.97 %	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBB	100 %	191130	Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant
TUBB2A	87.26 %	615101	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
TUBB2B	87.69 %	612850	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant
TUBB3	100 %	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
TUBB4A	100 %	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TUBG1	99.95 %	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
TUBGCP2	96.54 %	617817	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive
TUBGCP4	99.8 %	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	100 %	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TUFM	100 %	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
TUSC3	99.96 %	601385	Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive
TWIST1	100 %	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant
TWNK	100 %	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
U2AF2	99.98 %	191318	Developmental delay, dysmorphic facies, and brain anomalies, 620535 (3), Autosomal dominant
UBA5	99.95 %	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
UBAP2L	99.45 %	616472	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494 (3), Autosomal dominant
UBE2A	99.77 %	312180	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive
UBE3A	99.23 %	601623	Angelman syndrome, 105830 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
UBE3B	99.99 %	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
UBE3C	99.97 %	614454	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270 (3), Autosomal recessive
UBE4A	99.99 %	603753	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639 (3), Autosomal recessive
UBR1	99.93 %	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UBR7	99.96 %	613816	Li-Campeau syndrome, 619189 (3), Autosomal recessive
UBTF	99.99 %	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
UFC1	99.35 %	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
UFM1	99.25 %	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
UFSP2	99.74 %	611482	?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant; Developmental and epileptic encephalopathy 106, 620028 (3), Autosomal recessive
UGDH	99.77 %	603370	Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive
UGP2	99.85 %	191760	Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive
UMPS	99.9 %	613891	Orotic aciduria, 258900 (3), Autosomal recessive
UNC13A	99.99 %	609894	*No OMIM phenotypes*
UNC79	99.98 %	616884	*No OMIM phenotypes*
UNC80	98.04 %	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
UPB1	100 %	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
UPF1	99.97 %	601430	*No OMIM phenotypes*
UPF3B	99.26 %	300298	Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive
UQCRQ	99.96 %	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
UROC1	99.99 %	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
USP18	93.05 %	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
USP25	99.72 %	604736	*No OMIM phenotypes*
USP27X	99.99 %	300975	Intellectual developmental disorder, X-linked 105, 300984 (3), X-linked recessive
USP7	99.9 %	602519	Hao-Fountain syndrome, 616863 (3), Autosomal dominant
USP9X	99.84 %	300072	Intellectual developmental disorder, X-linked 99, 300919 (3), X-linked recessive; Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
VAMP1	100 %	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VAMP2	100 %	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
VARS1	99.99 %	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive
VARS2	99.98 %	612802	Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive
VCP	99.99 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
VLDLR	99.99 %	192977	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive
VPS11	99.99 %	608549	?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
VPS13B	99.9 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS16	100 %	608550	Dystonia 30, 619291 (3), Autosomal dominant
VPS33B	99.95 %	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
VPS37A	99.96 %	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
VPS41	99.92 %	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
VPS4A	99.98 %	609982	CIMDAG syndrome, 619273 (3), Autosomal dominant
VPS50	98.35 %	616465	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 (3), Autosomal recessive
VPS51	93 %	615738	Pontocerebellar hypoplasia, type 13, 618606 (3), Autosomal recessive
VPS53	100 %	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
VRK1	99.98 %	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 (3), Autosomal recessive
WAC	99.76 %	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
WARS1	99.97 %	191050	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317 (3), Autosomal recessive
WARS2	97.72 %	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
WASF1	99.86 %	605035	Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant
WASHC4	99.84 %	615748	Intellectual developmental disorder, autosomal recessive 43, 615817 (3), Autosomal recessive
WASHC5	99.98 %	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WBP4	99.95 %	604981	Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852 (3), Autosomal recessive
WDFY3	99.93 %	617485	?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant
WDR19	99.8 %	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR26	96.29 %	617424	Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant
WDR37	99.94 %	618586	Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant
WDR4	99.95 %	605924	Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive
WDR44	99.45 %	301070	*No OMIM phenotypes*
WDR45	99.99 %	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR45B	100 %	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
WDR47	95.08 %	615734	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
WDR5	100 %	609012	*No OMIM phenotypes*
WDR62	99.96 %	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
WDR73	99.92 %	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WDR81	100 %	614218	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
WDR83OS	100 %	618474	*No OMIM phenotypes*
WFS1	99.99 %	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
WIPI2	99.99 %	609225	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 (3), Autosomal recessive
WLS	99.6 %	611514	Zaki syndrome, 619648 (3), Autosomal recessive
WNK3	99.82 %	300358	Prieto syndrome, 309610 (3), X-linked recessive
WNT1	99.97 %	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3), Autosomal dominant; Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
WWOX	100 %	605131	Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XK	99.98 %	314850	McLeod syndrome, 300842 (3), X-linked
XPA	99.68 %	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPNPEP3	99.99 %	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XRCC4	99.89 %	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
XYLT1	99.98 %	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
XYLT2	99.97 %	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
YAP1	99.87 %	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433 (3), Autosomal dominant
YEATS2	99.96 %	613373	?Epilepsy, myoclonic, familial adult, 4, 615127 (3), Autosomal dominant
YIF1B	99.8 %	619109	Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive
YIPF5	99.99 %	611483	Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive
YME1L1	99.87 %	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
YWHAE	84.93 %	605066	*No OMIM phenotypes*
YWHAG	99.99 %	605356	Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant
YY1	100 %	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
ZBTB11	99.97 %	618181	Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive
ZBTB16	100 %	176797	Leukemia, acute promyelocytic, PL2F/RARA type (3)
ZBTB18	99.41 %	608433	Intellectual developmental disorder, autosomal dominant 22, 612337 (3), Autosomal dominant
ZBTB20	100 %	606025	Primrose syndrome, 259050 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ZBTB24	99.99 %	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
ZBTB47	96.86 %	619969	*No OMIM phenotypes*
ZBTB7A	100 %	605878	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769 (3), Autosomal dominant
ZC3H14	99.96 %	613279	Intellectual developmental disorder, autosomal recessive 56, 617125 (3), Autosomal recessive
ZC4H2	99.98 %	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
ZDHHC9	99.61 %	300646	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked
ZEB2	99.97 %	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
ZFHX3	99.96 %	104155	Prostate cancer, somatic, 176807 (3); {Atrial fibrillation 8, susceptibility to}, 613055 (3), Autosomal dominant; Spinocerebellar ataxia 4, 600223 (3), Autosomal dominant
ZFHX4	99.99 %	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
ZFX	94.74 %	314980	Intellectual developmental disorder, X-linked syndromic 37, 301118 (3), X-linked
ZFYVE26	100 %	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
ZIC1	99.99 %	600470	?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant
ZIC2	100 %	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
ZMIZ1	99.92 %	607159	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant
ZMYM2	99.88 %	602221	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant
ZMYM3	99.98 %	300061	Intellectual developmental disorder, X-linked 112, 301111 (3), X-linked recessive
ZMYND11	99.99 %	608668	Intellectual developmental disorder, autosomal dominant 30, 616083 (3), Autosomal dominant
ZMYND8	99.99 %	615713	*No OMIM phenotypes*
ZNF142	100 %	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive
ZNF148	99.85 %	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
ZNF292	99.91 %	616213	Intellectual developmental disorder, autosomal dominant 64, 619188 (3), Autosomal dominant
ZNF335	100 %	610827	Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive
ZNF407	99.98 %	615894	SIMHA syndrome, 619557 (3), Autosomal recessive
ZNF423	98.94 %	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive
ZNF462	99.99 %	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
ZNF526	100 %	614387	Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive
ZNF668	99.99 %	617103	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194 (3), Autosomal recessive
ZNF699	100 %	609571	DEGCAGS syndrome, 619488 (3), Autosomal recessive
ZNF711	99.43 %	314990	Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked
ZNFX1	99.99 %	618931	Immunodeficiency 91 and hyperinflammation, 619644 (3), Autosomal recessive
ZNHIT3	62.89 %	604500	PEHO syndrome, 260565 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ZNRF3	99.94 %	612062	*No OMIM phenotypes*
ZSCAN10	100 %	618365	Otofacial neurodevelopmental syndrome, 620910 (3), Autosomal dominant
ZSWIM6	98.94 %	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.