Whole Exome Sequencing
Gene package Intellectual disability, version 9.1, 31-1-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
A2ML1	{Otitis media, susceptibility to}, 166760	610627	66	100	**100**	**96**
AARS1	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	63	100	97	90
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	100	100	100	96
ABAT	GABA-transaminase deficiency, 613163	137150	65	100	100	95
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	86	84	78	75
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	108	100	100	100
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	109	100	100	100
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	74	100	100	98
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	159	100	100	100
ACAD9	Mitochondrial complex I deficiency, nuclear type 20, 611126	611103	95	100	100	100
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	146	100	98	94

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ACOT9	No OMIM phenotype	300862	36	100	90	67
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	103	100	100	99
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	149	100	100	100
ACSL4	Mental retardation 63, 300387	300157	54	100	98	88
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	192	100	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	188	100	100	100
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	73	100	100	99
ACY1	Aminoacylase 1 deficiency, 609924	104620	109	100	100	100
ADA2	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688	607575	96	100	100	99
ADAM22	?Epileptic encephalopathy, early infantile, 61, 617933	603709	66	100	99	94
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	90	100	100	100
ADAT3	Mental retardation 36, 615286	615302	152	100	100	100
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	120	100	100	100
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	53	100	93	80
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	75	100	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	89	100	100	96
AFF2	Mental retardation, FRAXE type, 309548	300806	57	100	99	92
AFF4	CHOPS syndrome, 616368	604417	72	100	99	93
AFG3L2	Spastic ataxia 5, 614487 Spinocerebellar ataxia 28, 610246	604581	77	99	95	90
AGA	Aspartylglucosaminuria, 208400	613228	73	100	100	96
AGAP2	No OMIM phenotype	605476	119	100	99	96
AGO1	No OMIM phenotype	606228	84	100	100	99
AGO2	No OMIM phenotype	606229	85	99	96	90
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	142	100	100	96
AGTPBP1	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276	606830	64	100	95	87
AGTR2	No OMIM phenotype	300034	55	100	100	100
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	151	96	96	96
AHDC1	Xia-Gibbs syndrome, 615829	615790	129	100	99	98
AHI1	Joubert syndrome 3, 608629	608894	67	100	98	90

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AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness 5, 300614	300169	55	100	96	82
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	78	100	100	96
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	124	100	100	100
AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500	164730	147	100	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	70	100	99	90
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	80	100	100	98
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	64	100	99	93
ALDH4A1	Hyperprolinemia, type II, 239510	606811	118	100	100	99
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	66	100	97	91
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	61	91	79	73
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	73	100	100	99
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	158	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	50	100	97	83
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	81	100	100	99
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	95	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	73	100	100	96
ALG8	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874	608103	62	100	100	94
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	64	100	100	94
ALMS1	Alstrom syndrome, 203800	606844	100	100	100	99
ALX1	?Frontonasal dysplasia 3, 613456	601527	78	100	100	100

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ALX4	{Craniosynostosis 5, susceptibility to}, 615529 Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597	605420	139	100	100	100
AMMECR1	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990	300195	48	100	88	75
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686	102771	142	100	100	100
AMT	Glycine encephalopathy, 605899	238310	130	100	100	100
ANK3	?Mental retardation, 37, 615493	600465	70	100	100	99
ANKEF1	No OMIM phenotype	No ID	80	100	100	97
ANKH	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	605145	91	100	100	99
ANKLE2	Microcephaly 16, primary, 616681	616062	102	100	99	95
ANKRD11	KBG syndrome, 148050	611192	112	100	98	96
ANO10	Spinocerebellar ataxia 10, 613728	613726	57	100	99	88
ANTXR1	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089	606410	60	99	97	92
AP1S1	MEDNIK syndrome, 609313	603531	84	100	100	96
AP1S2	Mental retardation syndromic 5, 304340	300629	40	100	87	62
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	66	100	98	87
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276	602166	100	100	100	98
AP4B1	Spastic paraplegia 47, 614066	607245	81	100	100	100
AP4E1	Spastic paraplegia 51, 613744 Stuttering, familial persistent, 1, 184450	607244	64	100	100	96
AP4M1	Spastic paraplegia 50, 612936	602296	130	100	100	99
AP4S1	Spastic paraplegia 52, 614067	607243	44	100	99	87
APC2	?Sotos syndrome 3, 617169	612034	128	100	97	95
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	76	100	99	92
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	83	100	100	96
ARG1	Argininemia, 207800	608313	75	100	100	98
ARHGAP31	Adams-Oliver syndrome 1, 100300	610911	103	100	100	99
ARHGAP4	No OMIM phenotype	300023	77	100	98	94
ARHGEF33	No OMIM phenotype	No ID	76	99	95	90
ARHGEF6	No OMIM phenotype	300267	47	100	97	83
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	50	100	99	85
ARID1A	Coffin-Siris syndrome 2, 614607	603024	121	100	99	97

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ARID1B	Coffin-Siris syndrome 1, 135900	614556	102	100	100	98
ARID2	Coffin-Siris syndrome 6, 617808	609539	76	100	100	96
ARL13B	Joubert syndrome 8, 612291	608922	63	100	100	94
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	43	96	95	83
ARNT2	?Webb-Dattani syndrome, 615926	606036	91	100	100	97
ARSA	Metachromatic leukodystrophy, 250100	607574	136	100	100	100
ARSL	Chondrodysplasia punctata recessive, 302950	300180	81	100	99	92
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly 2, 300215 Mental retardation 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	46	89	79	68
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	67	100	100	93
ASCL1	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880	100790	259	100	100	100
ASL	Argininosuccinic aciduria, 207900	608310	117	100	100	99
ASNS	Asparagine synthetase deficiency, 615574	108370	73	100	100	92
ASPA	Canavan disease, 271900	608034	55	100	99	91
ASPM	Microcephaly 5, primary, 608716	605481	71	100	100	97
ASS1	Citrullinemia, 215700	603470	115	100	98	90
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	107	100	98	98
ASXL2	Shashi-Pena syndrome, 617190	612991	88	100	99	96
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	69	99	99	97
ATAD3A	Harel-Yoon syndrome, 617183	612316	124	98	95	92
ATAD3B	No OMIM phenotype	612317	125	98	95	90
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	116	100	100	96
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	62	100	100	96
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 Dentatorubral-pallidoluysian atrophy, 125370	607462	155	100	100	99

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ATP1A2	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	130	100	100	100
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	144	100	100	100
ATP2A2	Acrokeratosis verruciformis, 101900 Darier disease, 124200	108740	99	100	100	100
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	46	100	91	64
ATP6V0A2	Cutis laxa, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	81	100	100	97
ATP6V1B2	Deafness, congenital, with onychodystrophy, 124480 Zimmermann-Laband syndrome 2, 616455	606939	81	100	100	99
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	49	100	98	84
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	69	100	99	94
ATPAF2	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	73	100	100	100
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600	601215	91	100	99	92
ATRIP	No OMIM phenotype	606605	108	100	100	97
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	40	100	94	78
AUH	3-methylglutaconic aciduria, type I, 250950	600529	94	100	100	98
AUTS2	Mental retardation 26, 615834	607270	122	100	100	98
AVPR2	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	300538	85	100	100	99
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	59	100	100	97
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	64	79	75	72
B3GLCT	Peters-plus syndrome, 261540	610308	67	100	99	85
B4GALNT1	Spastic paraplegia 26, 609195	601873	111	100	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	95	100	100	100

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B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	604327	126	100	100	98
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	154	100	100	100
BBS1	Bardet-Biedl syndrome 1, 209900	209901	126	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	66	100	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	59	100	100	99
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	77	100	100	97
BBS4	Bardet-Biedl syndrome 4, 615982	600374	76	100	100	95
BBS5	Bardet-Biedl syndrome 5, 615983	603650	65	100	98	89
BBS7	Bardet-Biedl syndrome 7, 615984	607590	64	100	99	95
BBS9	Bardet-Biedl syndrome 9, 615986	607968	59	96	95	90
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	83	100	100	97
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	160	100	100	100
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	64	100	99	94
BCL11A	Dias-Logan syndrome, 617101	606557	111	100	100	99
BCOR	Microphthalmia, syndromic 2, 300166	300485	78	100	99	94
BCORL1	Shukla-Vernon syndrome, 301029	300688	118	100	100	98
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	170	100	100	100
BLM	Bloom syndrome, 210900	604610	82	100	100	96
BRAF	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706	164757	68	100	100	94
BRAT1	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	125	100	100	100
BRF1	Cerebellofaciodental syndrome, 616202	604902	113	100	100	100
BRPF1	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333	602410	131	100	100	100
BRWD3	Mental retardation 93, 300659	300553	45	100	95	81

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BSCL2	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685	606158	101	100	100	100
BTD	Biotinidase deficiency, 253260	609019	89	100	100	100
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	70	100	99	94
C12orf4	Mental retardation 66, 618221	616082	81	100	100	95
C12orf57	Temtamy syndrome, 218340	615140	141	100	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, 615035	613541	78	100	100	100
C1orf167	No OMIM phenotype	No ID	98	100	97	94
C2CD3	Orofaciodigital syndrome XIV, 615948	615944	80	100	100	99
CA2	Osteopetrosis 3, with renal tubular acidosis, 259730	611492	99	100	100	100
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	175	100	100	100
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	79	100	100	94
CACNA1A	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086	601011	87	100	98	93
CACNA1C	Brugada syndrome 3, 611875 Long QT syndrome 8, 618447 Timothy syndrome, 601005	114205	123	100	100	99
CACNA2D1	No OMIM phenotype	114204	63	100	99	91
CACNG2	?Mental retardation 10, 614256	602911	107	100	100	99
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	121	100	100	100
CAMK2A	Mental retardation 53, 617798 ?Mental retardation 63, 618095	114078	78	100	99	95
CAMK2B	Mental retardation 54, 617799	607707	92	100	100	95
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	161	100	99	97
CAPN10	{Diabetes mellitus, noninsulin-dependent 1}, 601283	605286	107	100	100	100

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CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	51	100	96	80
CBL	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	78	100	100	100
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	127	100	100	100
CC2D1A	Mental retardation 3, 608443	610055	132	100	100	100
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	66	100	100	95
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	87	100	99	95
CCDC13	No OMIM phenotype	No ID	85	100	100	100
CCDC14	No OMIM phenotype	617147	74	100	100	96
CCDC174	Hypotonia, infantile, with psychomotor retardation, 616816	616735	68	100	97	84
CCDC22	Ritscher-Schinzel syndrome 2, 300963	300859	81	100	94	92
CCDC78	?Centronuclear myopathy 4, 614807	614666	123	100	100	100
CCDC88C	Hydrocephalus, congenital, 1, 236600 ?Spinocerebellar ataxia 40, 616053	611204	104	100	100	97
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	123833	107	100	100	99
CDC5L	No OMIM phenotype	602868	54	100	99	90
CDH15	Mental retardation 3, 612580	114019	125	100	100	100
CDK16	No OMIM phenotype	311550	75	100	100	97
CDK5	?Lissencephaly 7 with cerebellar hypoplasia, 616342	123831	118	100	100	100
CDK5RAP2	Microcephaly 3, primary, 604804	608201	69	100	99	94
CDK6	?Microcephaly 12, primary, 616080	603368	78	100	100	96
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	58	100	97	87
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	74	90	83	76
CDON	Holoprosencephaly 11, 614226	608707	89	100	100	99
CENPJ	Microcephaly 6, primary, 608393 ?Seckel syndrome 4, 613676	609279	74	100	100	98
CEP104	Joubert syndrome 25, 616781	616690	71	100	98	93
CEP135	Microcephaly 8, primary, 614673	611423	83	100	99	91

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CEP152	Microcephaly 9, primary, 614852 Seckel syndrome 5, 613823	613529	67	100	98	93
CEP290	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189	610142	71	100	98	89
CEP41	Joubert syndrome 15, 614464	610523	69	100	100	94
CEP63	?Seckel syndrome 6, 614728	614724	72	100	99	92
CEP89	No OMIM phenotype	615470	68	100	98	91
CERT1	Mental retardation 34, 616351	604677	No coverage data			
CHAMP1	Mental retardation 40, 616579	616327	128	100	100	100
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	72	100	100	96
CHD3	Snijders Blok-Campeau syndrome, 618205	602120	86	98	96	91
CHD4	Sifrim-Hitz-Weiss syndrome, 617159	603277	70	100	100	98
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	87	100	100	97
CHD8	{Autism, susceptibility to, 18}, 615032	610528	85	100	100	98
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	111	100	100	100
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	102	100	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	138	100	100	96
CIC	Mental retardation 45, 617600	612082	134	100	100	99
CIT	Microcephaly 17, primary, 617090	605629	91	100	100	97
CKAP2L	Filippi syndrome, 272440	616174	81	100	100	100
CLCN4	Raynaud-Claes syndrome, 300114	302910	80	100	100	98
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	138	100	100	100
CLIC2	?Mental retardation, syndromic 32, 300886	300138	40	100	90	69
CLIP1	No OMIM phenotype	179838	77	100	99	93
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	111	100	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	70	100	100	100
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	120	100	100	99

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CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	129	100	100	100
CLP1	Pontocerebellar hypoplasia, type 10, 615803	608757	99	100	100	100
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	114	100	100	100
CLTC	Mental retardation 56, 617854	118955	65	100	98	94
CNKSR2	Mental retardation, syndromic, Houge type, 301008	300724	56	100	97	87
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	161	100	100	99
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	73	100	100	99
COASY	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266	609855	139	100	100	100
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	96	100	100	98
COG4	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150	606976	81	100	100	99
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	66	100	99	91
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	65	100	99	90
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	83	100	100	98
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	125	100	100	100
COL11A1	?Deafness 37, 618533 Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	62	100	99	93
COL12A1	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470	120320	71	100	100	96
COL18A1	Knobloch syndrome, type 1, 267750	120328	143	100	100	97
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000	120130	88	100	100	97
COL4A2	Brain small vessel disease 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519	120090	102	100	100	99

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COLEC11	3MC syndrome 2, 265050	612502	168	100	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	66	100	100	94
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276	612898	110	100	100	100
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	129	100	100	100
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	105	100	100	100
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	139	100	100	99
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	65	100	98	92
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	102	100	100	100
CPLANE1	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	71	100	99	95
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371	608307	61	100	100	96
CRADD	Mental retardation 34, with variant lissencephaly, 614499	603454	139	100	100	100
CRB2	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	609720	115	100	100	100
CRBN	Mental retardation 2, 607417	609262	67	100	99	91
CREBBP	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849	600140	85	100	99	94
CRLF1	Cold-induced sweating syndrome 1, 272430	604237	105	96	91	89
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	81	100	99	93
CSNK2A1	Okur-Chung neurodevelopmental syndrome, 617062	115440	69	100	95	86
CSPP1	Joubert syndrome 21, 615636	611654	83	100	100	97
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	84	100	100	100
CTBP1	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915	602618	96	95	85	82
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	100	100	100	100
CTCF	Mental retardation 21, 615502	604167	98	100	100	100
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	125	98	89	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CTNNB1	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	62	100	100	97
CTNND1	Blepharocheilodontic syndrome 2, 617681	601045	74	100	100	96
CTNND2	No OMIM phenotype	604275	86	97	94	90
CTSA	Galactosialidosis, 256540	613111	124	100	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	137	100	100	100
CTTNBP2	No OMIM phenotype	609772	96	100	100	97
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	82	100	100	96
CUL3	Pseudohypoaldosteronism, type IIE, 614496	603136	73	100	97	88
CUL4B	Mental retardation, syndromic 15 (Cabezas type), 300354	300304	52	100	96	82
CWF19L1	Spinocerebellar ataxia 17, 616127	616120	59	100	100	92
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	127	100	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	131	100	100	100
CYP2U1	Spastic paraplegia 56, 615030	610670	66	100	100	96
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	140	100	100	100
DAB1	Spinocerebellar ataxia 37, 615945	603448	70	100	100	97
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	128239	164	100	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	63	100	100	97
DBT	Maple syrup urine disease, type II, 248600	248610	115	100	100	99
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	75	100	100	94
DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600	120470	70	100	100	98
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	128	100	100	100
DCPS	Al-Raqad syndrome, 616459	610534	121	100	100	100

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DCX	Lissencephaly, 300067 Subcortical laminal heterotopia, 300067	300121	57	100	98	92
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	84	100	99	92
DDHD2	Spastic paraplegia 54, 615033	615003	76	100	100	97
DDX11	Warsaw breakage syndrome, 613398	601150	254	100	100	100
DDX3X	Mental retardation 102, 300958 dominant	300160	76	100	100	98
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation 24, 615828	602635	86	100	97	90
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281	617278	64	100	99	94
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	83	100	100	97
DHCR24	Desmosterolosis, 602398	606418	119	100	100	100
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	112	100	100	100
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	76	100	100	98
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	85	100	100	98
DIAPH1	Deafness 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	84	100	99	93
DIP2B	Mental retardation, FRA12A type, 136630	611379	65	100	100	97
DKC1	Dyskeratosis congenita, 305000	300126	53	100	98	89
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	77	100	100	98
DLG3	Mental retardation 90, 300850	300189	61	100	97	88
DLG4	No OMIM phenotype	602887	118	100	100	100
DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	46	100	96	81
DMPK	Myotonic dystrophy 1, 160900	605377	117	100	100	99
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	69	100	95	88
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	75	100	100	96
DNM1	Epileptic encephalopathy, early infantile, 31, 616346	602377	138	100	100	95
DNMT3A	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879	602769	117	100	100	99
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	115	100	100	99
DOCK6	Adams-Oliver syndrome 2, 614219	614194	108	100	98	96
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859	615730	56	100	99	93
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	74	100	99	94

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DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	136	100	100	100
DONSON	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230	611428	45	100	91	75
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	76	100	100	100
DPF2	Coffin-Siris syndrome 7, 618027	601671	82	100	99	94
DPH1	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901	603527	126	100	100	100
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	79	94	89	85
DPP6	Mental retardation 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956	126141	82	100	97	90
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	66	100	99	95
DPYS	Dihydropyrimidinuria, 222748	613326	75	100	100	98
DST	Epidermolysis bullosa simplex 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653	113810	67	100	99	95
DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	67	100	98	88
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	96	100	100	99
DYNC1I2	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492	603331	36	93	72	54
DYRK1A	Mental retardation 7, 614104	600855	75	100	100	98
EBP	Chondrodysplasia punctata dominant, 302960 MEND syndrome, 300960	300205	93	100	100	100
EDC3	?Mental retardation 50, 616460	609842	115	100	100	100
EDNRB	ABCD syndrome, 600501 {Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580	131244	96	100	100	100
EDRF1	No OMIM phenotype	No ID	64	100	99	95
EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation 38, 616393	602959	169	100	100	100
EFCAB1	No OMIM phenotype	No ID	67	100	100	94
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536	603892	85	100	100	99
EHMT1	Kleefstra syndrome 1, 610253	607001	126	99	99	99
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	72	100	99	94

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EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305	608546	68	100	100	97
EIF4G1	{Parkinson disease 18}, 614251	600495	100	100	100	100
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731	605367	87	100	100	98
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	71	100	100	98
ELP2	Mental retardation 58, 617270	616054	75	100	100	99
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	117	100	100	99
EMG1	Bowen-Conradi syndrome, 211180	611531	78	100	100	100
EML1	Band heterotopia, 600348	602033	76	100	99	94
EMX2	Schizencephaly, 269160	600035	164	100	100	100
ENTPD1	Spastic paraplegia 64, 615683	601752	68	100	100	96
EOMES	No OMIM phenotype	604615	89	100	100	100
EP300	Colorectal cancer, somatic, 114500 Menke-Hennekam syndrome 2, 618333 Rubinstein-Taybi syndrome 2, 613684	602700	109	100	100	97
EPB41L1	?Mental retardation 11, 614257	602879	116	100	100	99
EPG5	Vici syndrome, 242840	615068	69	100	100	97
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	71	100	100	94
ERCC2	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	97	100	99	98
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	133510	82	100	100	99
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	133530	85	100	100	98
ERCC6	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630	609413	88	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	81	100	99	91
ERLIN2	Spastic paraplegia 18, 611225	611605	65	100	100	95
ERMARD	?Periventricular nodular heterotopia 6, 615544	615532	67	100	100	96
ESCO2	Roberts syndrome, 268300 SC phocomelia syndrome, 269000	609353	58	100	100	95
ETFB	Glutaric acidemia IIB, 231680	130410	106	100	100	100
ETHE1	Ethylmalonic encephalopathy, 602473	608451	113	100	100	98
EXOSC2	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	602238	69	100	100	97
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	114	100	100	96
EZH2	Weaver syndrome, 277590	601573	79	100	100	97
FA2H	Spastic paraplegia 35, 612319	611026	83	100	99	91
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	63	100	100	98
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154	616107	74	100	100	96
FAT2	Spinocerebellar ataxia 45, 617769	604269	95	100	100	99
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	90	100	100	99
FBN1	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328	134797	186	100	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	74	100	100	97
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	605654	67	100	100	98
FBXO31	?Mental retardation 45, 615979	609102	103	100	99	96
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation syndromic 16, 305400	300546	71	100	98	95
FGF12	Epileptic encephalopathy, early infantile, 47, 617166	601513	52	100	97	85
FGF14	Spinocerebellar ataxia 27, 609307	601515	68	100	100	98

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FGFR1	Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440	136350	102	100	100	98
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579	176943	74	100	100	95
FGFR3	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601	134934	122	100	100	98

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FGL1	No OMIM phenotype	605776	55	100	99	90
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	82	99	94	87
FIBP	Thauvin-Robinet-Faivre syndrome, 617107	608296	104	100	100	100
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	59	100	99	94
FIGN	No OMIM phenotype	605295	87	100	100	100
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	143	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	85	100	100	100
FLNA	Cardiac valvular dysplasia, 314400 Congenital short bowel syndrome, 300048 ?FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	110	100	100	100
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	98	100	100	96
FMN2	Mental retardation 47, 616193	606373	102	100	94	89
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	41	100	96	76
FOXG1	Rett syndrome, congenital variant, 613454	164874	127	97	90	84
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	80	100	100	98
FOXP2	Speech-language disorder-1, 602081	605317	72	100	100	98
FRAS1	Fraser syndrome 1, 219000	607830	79	100	100	97

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FREM2	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666	608945	104	100	100	100
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	616305	86	100	97	90
FRMPD4	Mental retardation 104, 300983	300838	69	100	99	95
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981	604574	50	85	79	70
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	96	99	96	93
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	90	100	100	99
FTSJ1	Mental retardation 9/44, 309549	300499	91	100	100	93
FUCA1	Fucosidosis, 230000	612280	83	100	100	98
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	76	100	100	100
GABRB1	Epileptic encephalopathy, early infantile, 45, 617153	137190	86	100	100	97
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	95	100	100	98
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	86	100	100	99
GALE	Galactose epimerase deficiency, 230350	606953	116	100	100	100
GALT	Galactosemia, 230400	606999	151	100	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	85	100	99	95
GAS6	No OMIM phenotype	600441	94	100	96	93
GATAD2B	Mental retardation 18, 615074	614998	67	100	100	99
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	65	100	100	98
GCDH	Glutaricaciduria, type I, 231670	608801	112	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	52	100	100	93
GCSH	?Glycine encephalopathy, 605899	238330	93	100	89	62
GDI1	Mental retardation 41, 300849	300104	97	100	100	100
GFAP	Alexander disease, 203450	137780	81	100	100	99
GFM2	Combined oxidative phosphorylation deficiency 39, 618397	606544	64	100	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	101	100	100	100
GJB1	Charcot-Marie-Tooth neuropathy dominant, 1, 302800	304040	91	100	100	99
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, 613206	608803	85	96	86	76
GK	Glycerol kinase deficiency, 307030	300474	46	99	88	66
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	105	100	100	98
GLDC	Glycine encephalopathy, 605899	238300	67	100	98	91
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	163	100	100	99
GLI3	Greig cephalopolysyndactyly syndrome, 175700 {Hypothalamic hamartomas, somatic}, 241800 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700	165240	110	100	100	99
GLYCTK	D-glyceric aciduria, 220120	610516	136	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	94	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	114	100	100	100
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352	615320	189	100	100	100
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493	139311	98	100	100	99
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	600998	94	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463	139320	180	100	100	97
GNB1	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation 42, 616973	139380	93	100	100	96
GNB5	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182	604447	66	100	99	94
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	78	100	100	96
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	63	100	99	95
GNS	Mucopolysaccharidosis type IIID, 252940	607664	68	100	100	98
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	53	100	99	91
GPHN	Molybdenum cofactor deficiency C, 615501	603930	68	100	100	98
GPT2	Mental retardation 49, 616281	138210	97	100	96	85
GRIA3	Mental retardation 94, 300699	305915	51	100	98	88
GRID2	Spinocerebellar ataxia 18, 616204	602368	78	100	100	99
GRIK2	Mental retardation, 6, 611092	138244	94	100	100	98
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, 617820	138249	132	100	100	99
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	103	100	100	100
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	122	100	100	99
GRIN3B	No OMIM phenotype	606651	137	91	86	80
GRIP1	Fraser syndrome 3, 617667	604597	84	100	100	99
GRM1	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia 13, 614831	604473	136	100	100	99
GSE1	No OMIM phenotype	616886	116	100	100	100
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	89	100	100	99

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GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	53	100	100	100
GTPBP3	Combined oxidative phosphorylation deficiency 23, 616198	608536	148	100	100	100
GUCY2F	No OMIM phenotype	300041	44	100	96	84
GUSB	Mucopolysaccharidosis VII, 253220	611499	106	100	100	97
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data			
H3-3A	No OMIM phenotype	601128	No coverage data			
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures, 616756	610876	80	100	100	94
HAX1	Neutropenia, severe congenital 3, 610738	605998	99	100	100	99
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	45	100	99	84
HCFC1	Mental retardation 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	79	100	97	92
HCN1	Epileptic encephalopathy, early infantile, 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482	602780	79	100	100	98
HDAC4	No OMIM phenotype	605314	112	100	100	100
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	300272	85	100	100	97
HDAC8	Cornelia de Lange syndrome 5, 300882	300269	47	100	100	91
HECTD1	No OMIM phenotype	No ID	58	100	97	88
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	617245	74	100	97	90
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	97	100	100	99
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011	605109	66	100	99	94
HERC2	Mental retardation 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	102	100	99	96
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230	601802	71	100	100	95
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	86	100	100	98
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	119	100	100	97
HIVEP2	Mental retardation 43, 616977	143054	102	100	100	98
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	98	100	100	99

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HMGCL	HMG-CoA lyase deficiency, 246450	613898	94	100	100	100
HNMT	{Asthma, susceptibility to}, 600807 Mental retardation 51, 616739	605238	75	100	100	99
HNRNPH2	Mental retardation, syndromic, Bain type, 300986	300610	79	100	100	100
HNRNPK	Au-Kline syndrome, 616580	600712	46	96	84	66
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391	602869	81	100	100	95
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	126	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	107	100	100	99
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	43	100	99	81
HRAS	Bladder cancer, somatic, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Thyroid carcinoma, follicular, somatic, 188470	190020	179	100	100	100
HSD17B10	HSD10 mitochondrial disease, 300438	300256	76	100	100	100
HSPA9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854	600548	70	100	99	93
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, 605280	118190	65	100	99	85
HUWE1	Mental retardation syndromic, Turner type, 309590	300697	53	100	96	83
IARS1	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093	600709	51	100	98	88
IBA57	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, 616451	615316	124	100	100	97
IDS	Mucopolysaccharidosis II, 309900	300823	74	100	99	95
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	138	100	97	91
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	83	100	100	77
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	86	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	75	100	100	96
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895	605489	47	99	94	82
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	69	100	100	90
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	71	100	100	100
IKBKG	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	24	38	33	31
IL1RAPL1	Mental retardation 21/34, 300143	300206	52	100	98	88
IMPA1	Mental retardation 59, 617323	602064	53	99	91	73
INPP5B	No OMIM phenotype	147264	94	100	100	98
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	113	100	100	97
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404	607875	72	100	100	98
INTS1	No OMIM phenotype	611345	106	100	99	96
INTS8	No OMIM phenotype	611351	50	100	98	84
IQSEC2	Mental retardation 1/78, 309530	300522	66	98	92	85
ISG15	Immunodeficiency 38, 616126	147571	144	100	100	100
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	600536	117	100	97	96
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	90	100	100	98
ITPR2	?Anhidrosis, isolated, with normal sweat glands, 106190	600144	57	100	96	84
ITSN1	No OMIM phenotype	602442	66	100	97	91
IVD	Isovaleric acidemia, 243500	607036	107	100	100	98
JAG1	Alagille syndrome 1, 118450 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Tetralogy of Fallot, 187500	601920	96	100	100	96
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	70	100	100	97
JMJD1C	No OMIM phenotype	604503	68	100	99	94
KALRN	{Coronary heart disease, susceptibility to, 5}, 608901	604605	90	100	99	96
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900	607704	105	100	100	99

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KANSL1	Koolen-De Vries syndrome, 610443	612452	85	100	100	97
KAT6A	Mental retardation 32, 616268	601408	95	100	100	99
KAT6B	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736	605880	107	100	100	98
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	141	100	100	100
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366	176262	108	100	100	100
KCNA4	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284	176266	91	100	100	100
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056	600397	123	100	100	100
KCNC3	Spinocerebellar ataxia 13, 605259	176264	91	94	78	64
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500	603305	114	100	100	98
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	171	100	100	100
KCNJ11	Diabetes mellitus, transient neonatal, 3, 610582 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329	600937	128	100	100	100
KCNJ6	Keppen-Lubinsky syndrome, 614098	600877	98	100	100	100
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292	605874	129	100	100	100
KCNMA1	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446	600150	88	100	100	96
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data			
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200	602235	131	100	100	100
KCNQ3	Seizures, benign neonatal, 2, 121201	602232	108	100	100	96
KCNQ5	Mental retardation 46, 617601	607357	85	100	98	95
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959	608167	120	100	99	99
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	159	100	100	100
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features, 616728	609132	61	100	100	93
KDM4B	No OMIM phenotype	609765	121	100	99	98
KDM5C	Mental retardation, syndromic, Claes-Jensen type, 300534	314690	88	100	99	96
KDM6A	Kabuki syndrome 2, 300867	300128	52	100	95	80

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KDM6B	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505	611577	132	100	98	95
KDSR	Erythrokeratodermia variabilis et progressiva 4, 617526	136440	65	100	100	96
KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	610178	72	100	98	94
KIAA1109	Alkuraya-Kucinskas syndrome, 617822	611565	78	100	100	97
KIAA1586	No OMIM phenotype	No ID	61	96	95	93
KIDINS220	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296	615759	59	100	98	90
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	73	100	99	93
KIF1A	Mental retardation 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, 610357	601255	101	100	100	98
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	84	100	99	90
KIF4A	?Mental retardation 100, 300923	300521	48	100	96	79
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282	604593	70	100	99	93
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	107	98	96	93
KIFBP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	76	100	100	98
KIRREL3	No OMIM phenotype	607761	114	100	100	100
KLHL15	Mental retardation 103, 300982	300980	49	100	99	90
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	71	100	100	98
KMT2B	Dystonia 28, childhood-onset, 617284	606834	141	97	95	94
KMT2C	Kleefstra syndrome 2, 617768	606833	82	100	99	96
KMT2D	Kabuki syndrome 1, 147920	602113	114	100	100	99
KMT2E	O'Donnell-Luria-Rodan syndrome, 618512	608444	82	100	100	97
KMT5B	Mental retardation 51, 617788	610881	96	100	100	99
KNL1	Microcephaly 4, primary, 604321	609173	60	100	99	95
KPTN	Mental retardation 41, 615637	615620	150	100	100	100

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KRAS	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Oculoectodermal syndrome, somatic, 600268 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	190070	83	100	100	87
KRBOX4	No OMIM phenotype	300585	66	100	100	95
L1CAM	CRASH syndrome, 303350 Corpus callosum, partial agenesis of, 304100 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 MASA syndrome, 303350	308840	101	100	100	100
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	74	100	100	97
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	87	100	100	97
LAMA2	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle 23, 618138	156225	71	100	99	96
LAMC1	No OMIM phenotype	150290	83	100	100	97
LAMC3	Cortical malformations, occipital, 614115	604349	130	100	100	99
LAMP2	Danon disease, 300257	309060	44	100	95	77
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840	603590	97	100	100	99
LARP7	Alazami syndrome, 615071	612026	67	100	99	92
LAS1L	Wilson-Turner syndrome, 309585	300964	71	100	100	97
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462	607031	78	100	100	95
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	71	100	100	100
LINS1	Mental retardation 27, 614340	610350	57	100	100	94
LMAN2L	?Mental retardation, 52, 616887	609552	77	100	98	94

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LONP1	CODAS syndrome, 600373	605490	131	100	100	100
LRP2	Donnai-Barrow syndrome, 222448	600073	69	100	100	96
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	60	100	99	91
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	79	100	100	93
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	604357	170	100	100	100
MACF1	Lissencephaly 9 with complex brainstem malformation, 618325	608271	68	100	99	94
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	80	84	80	76
MAGEC3	No OMIM phenotype	300469	66	100	96	88
MAGEL2	Schaaf-Yang syndrome, 615547	605283	88	100	98	93
MAGT1	Immunodeficiency, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	47	100	99	85
MAN1B1	Mental retardation 15, 614202	604346	122	100	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	115	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	85	100	100	95
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	49	100	99	92
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	79	100	100	96
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	117	100	100	95
MAPRE2	Symmetric circumferential skin creases, congenital, 2, 616734	605789	81	100	98	90
MASP1	3MC syndrome 1, 257920	600521	112	100	100	99
MAST1	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273	612256	143	100	100	100
MAT1A	Hypermethioninemia, persistent, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, 250850	610550	107	100	100	100
MBD5	Mental retardation 1, 156200	611472	81	100	100	99
MBOAT7	Mental retardation 57, 617188	606048	103	100	100	100
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, 308800 ?Olmsted syndrome, 300918 Osteogenesis imperfecta, type XIX, 301014	300294	46	100	99	90
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	74	100	100	94
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	64	100	100	96
MCM3AP	Peripheral neuropathy, with or without impaired intellectual development, 618124	603294	81	100	99	96
MCOLN1	Mucolipidosis IV, 252650	605248	129	100	100	100
MCOLN3	No OMIM phenotype	607400	45	100	94	83
MCPH1	Microcephaly 1, primary, 251200	607117	85	94	94	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MDH2	Epileptic encephalopathy, early infantile, 51, 617339	154100	103	100	100	100
MECP2	{Autism susceptibility 3}, 300496 Encephalopathy, neonatal severe, 300673 Mental retardation syndromic, Lubs type, 300260 Mental retardation, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	113	100	100	96
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	97	100	100	99
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, 300895 Opitz-Kaveggia syndrome, 305450	300188	68	100	100	97
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808	608771	73	100	100	97
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	94	100	100	99
MED23	Mental retardation 18, 614249	605042	63	100	100	96
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589	610197	137	100	100	99
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	93	100	99	96
MEIS2	Cleft palate, cardiac defects, and mental retardation, 600987	601740	86	100	100	99
METTL23	Mental retardation 44, 615942	615262	66	100	100	98
MFSD2A	Microcephaly 15, primary, 616486	614397	87	100	100	98
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	68	100	100	95
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	104	100	100	100
MICU1	Myopathy with extrapyramidal signs, 615673	605084	52	100	98	82
MID1	Opitz GBBB syndrome, type I, 300000	300552	89	100	100	93
MID2	?Mental retardation 101, 300928	300204	57	100	96	82
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	80	100	100	100
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	117	100	100	99
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	79	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	65	100	96	92
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	78	100	100	98
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	133	100	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	63	100	100	90
MMUT	Methylmalonic aciduria, mut(0) type, 251000	609058	75	100	100	95
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	105	100	100	99
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	67	100	100	96
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	130	100	100	100
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	87	100	100	99
MPDZ	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219	603785	69	100	99	95
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	56	100	100	100
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582	607118	64	100	100	90
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117	605810	75	100	100	96
MSL2	No OMIM phenotype	614802	78	100	100	99
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248	611766	78	100	100	90
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	99	100	100	99
MTMR9	No OMIM phenotype	606260	81	100	100	97
MTOR	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638	601231	96	100	100	97
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	80	100	100	97
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	87	100	100	98
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	100	100	100	100
MYCN	Feingold syndrome 1, 164280	164840	169	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MYH9	Deafness 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100	160775	125	100	100	99
MYO5A	Griscelli syndrome, type 1, 214450	160777	71	100	99	94
MYT1L	Mental retardation 39, 616521	613084	95	100	100	97
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	89	100	100	94
NAA15	Mental retardation 50, 617787	608000	79	100	100	95
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393	610672	164	100	100	100
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	118	100	100	100
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	112	100	97	93
NALCN	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	611549	69	100	100	97
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	65	100	99	94
NARS2	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness 94, 618434	612803	55	99	97	92
NAT10	No OMIM phenotype	609221	86	100	99	96
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	68	100	99	88
NCOR2	No OMIM phenotype	600848	86	99	94	86
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	109	100	100	100
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	77	100	100	100
NDST1	Mental retardation 46, 616116	600853	149	100	100	100
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12, 301020	300078	110	100	100	100
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14, 618236	612638	117	100	100	100
NDUFA12	?Mitochondrial complex I deficiency, nuclear type 23, 618244	614530	72	100	100	100
NDUFA2	?Mitochondrial complex I deficiency, nuclear type 13, 618235	602137	104	100	100	100
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18, 618240	612911	130	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16, 618238	612360	70	100	100	96
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5, 618226	157655	69	100	100	94
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6, 618228	602985	79	100	100	99
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8, 618230	603846	112	100	100	100
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1, 252010	602694	83	100	100	99
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9, 618232	603848	70	100	100	100
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3, 618224	601825	130	100	100	98
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2, 618222	602141	140	100	100	100
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4, 618225	161015	129	100	100	100
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7, 618229	600532	48	100	95	80
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	82	100	100	100
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	600644	112	100	100	100
NEDD4L	Periventricular nodular heterotopia 7, 617201	606384	71	100	99	94
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	155	100	100	100
NEXMIF	Mental retardation 98, 300912	300524	48	100	100	95
NF1	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	55	97	90	79
NFATC1	No OMIM phenotype	600489	167	100	100	100
NFE2L2	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744	600492	67	100	99	94
NFIA	Brain malformations with or without urinary tract defects, 613735	600727	87	100	100	98
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	154	100	100	100
NGLY1	Congenital disorder of deglycosylation, 615273	610661	78	100	100	97
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	57	100	96	90
NID1	No OMIM phenotype	131390	114	100	100	100
NIN	?Seckel syndrome 7, 614851	608684	79	100	99	95
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	65	100	98	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550	600635	95	100	100	100
NLGN3	{Asperger syndrome susceptibility 1}, 300494 {Autism susceptibility 1}, 300425	300336	100	100	100	98
NLGN4X	{Asperger syndrome susceptibility 2}, 300497 {Autism susceptibility 2}, 300495 Mental retardation, 300495	300427	179	100	100	100
NLRP3	CINCA syndrome, 607115 Deafness 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900	606416	125	100	100	100
NONO	Mental retardation, syndromic 34, 300967	300084	50	98	91	76
NOVA2	No OMIM phenotype	601991	111	97	93	88
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	87	100	100	99
NPC2	Niemann-pick disease, type C2, 607625	601015	95	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	58	100	98	88
NPHS1	Nephrotic syndrome, type 1, 256300	602716	102	100	100	100
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118	600928	85	100	99	95
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	197	100	100	100
NRAS	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470	164790	63	100	100	98
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	104	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NSD1	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	77	100	100	98
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	68	100	100	93
NSUN2	Mental retardation 5, 611091	610916	85	100	99	92
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	119	100	100	99
NUP62	Striatonigral degeneration, infantile, 271930	605815	107	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	64	100	96	85
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	61	96	84	79
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	43	100	95	80
ODC1	{Colonic adenoma recurrence, reduced risk of}, 114500	165640	68	100	100	100
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	43	100	95	75
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	52	100	96	84
ORC1	Meier-Gorlin syndrome 1, 224690	601902	86	100	100	97
OTC	Ornithine transcarbamylase deficiency, 311250	300461	58	100	98	85
P2RX6	No OMIM phenotype	608077	119	100	95	91
PACS1	Schuurs-Hoeijmakers syndrome, 615009	607492	92	100	100	97
PACS2	Epileptic encephalopathy, early infantile, 66, 618067	610423	140	100	99	96
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	82	100	96	90
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	68	100	99	95
PAK3	Mental retardation 30/47, 300558	300142	48	100	96	83
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	81	100	100	99
PANX1	Oocyte maturation defect 7, 618550	608420	73	100	99	93
PAX1	?Otofaciocervical syndrome 2, 615560	167411	202	100	90	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	73	100	100	96
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	167415	111	100	100	97
PBX1	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641	176310	72	100	100	98
PC	Pyruvate carboxylase deficiency, 266150	608786	141	100	100	100
PCCA	Propionicacidemia, 606054	232000	68	100	100	95
PCCB	Propionicacidemia, 606054	232050	72	100	98	95
PCDH12	Microcephaly, seizures, spasticity, and brain calcification, 251280	605622	144	100	100	100
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	113	100	100	97
PCGF2	Turnpenny-Fry syndrome, 618371	600346	88	100	99	96
PCLO	?Pontocerebellar hypoplasia, type 3, 608027	604918	80	100	99	98
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	121	100	100	99
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613	600129	70	100	97	94
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	49	100	94	84
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	87	100	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	60	100	93	85
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	75	100	97	87
PEPD	Prolidase deficiency, 170100	613230	100	100	100	98
PET100	Mitochondrial complex IV deficiency, 220110	614770	85	92	66	66
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	62	100	99	94
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	100	100	100	97
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	137	100	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	67	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	69	100	100	99
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	134	100	96	94
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	67	100	100	98
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	63	100	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	118	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	57	100	100	95
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	111	100	100	100
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	107	100	99	95
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	57	100	100	95
PGAP1	Mental retardation 42, 615802	611655	65	100	99	90
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	147	100	100	100
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	115	100	100	100
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	47	100	100	89
PHC1	?Microcephaly 11, primary, 615414	602978	122	100	97	94
PHF23	No OMIM phenotype	612910	118	100	100	100
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	51	100	96	82
PHF8	Mental retardation syndrome, Siderius type, 300263	300560	61	100	98	87
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	138	100	100	100
PHIP	Developmental delay, intellectual disability, obesity, and dysmorphism, 617991	612870	69	100	98	90
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	600286	114	100	99	96
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	67	100	100	97
PIGC	Glycosylphosphatidylinositol biosynthesis defect 16, 617816	601730	149	100	100	100
PIGG	Mental retardation 53, 616917	616918	100	100	100	98

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PIGL	CHIME syndrome, 280000	605947	98	100	100	100
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	63	100	99	89
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	126	100	100	100
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	138	100	100	100
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	96	100	100	100
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11, 616025	610275	85	100	100	100
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	44	100	100	94
PIK3CA	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000	171834	85	100	99	96
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	89	95	93	90
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	118	100	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	60	100	99	94
PLK4	Microcephaly and chorioretinopathy, 2, 616171	605031	62	100	99	91
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	93	100	99	97
PLXNA3	No OMIM phenotype	300022	99	98	97	95
PLXND1	No OMIM phenotype	604282	99	100	99	96
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	72	100	100	97
PMPCA	Spinocerebellar ataxia 2, 213200	613036	133	100	100	100
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	104	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	70	100	100	97
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	90	100	100	100
POGZ	White-Sutton syndrome, 616364	614787	88	100	100	97
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	114	100	100	99
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090	614258	84	100	100	98
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	76	100	99	93
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123	606822	93	100	100	99
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135	614828	154	100	100	100
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094	615247	95	100	100	100
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	103	100	100	100
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	87	100	100	99
PORCN	Focal dermal hypoplasia, 305600	300651	102	100	100	98
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	83	100	100	100
POU3F3	No OMIM phenotype	602480	99	77	68	62
PPFIA4	No OMIM phenotype	603145	97	100	99	95
PPM1D	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450	605100	88	100	100	99
PPOX	Porphyria variegata, 176200	600923	106	100	100	100
PPP1CB	Noonan syndrome-like disorder with loose anagen hair 2, 617506	600590	54	100	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2, 616817	613257	90	100	100	98
PPP2CA	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354	176915	74	100	100	93
PPP2R1A	Mental retardation 36, 616362	605983	130	100	100	100
PPP2R5B	No OMIM phenotype	601644	94	100	100	100
PPP2R5C	No OMIM phenotype	601645	59	97	89	78
PPP2R5D	Mental retardation 35, 616355	601646	99	100	100	98
PPP3CA	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Epileptic encephalopathy, infantile or early childhood, 1, 617711	114105	76	100	99	90
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	84	100	100	100
PQBP1	Renpenning syndrome, 309500	300463	100	100	100	100
PRIM1	No OMIM phenotype	176635	51	100	95	79
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489	188830	86	100	100	99
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157	610087	98	100	100	98
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	98	100	95	86
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	50	100	99	90
PRR12	No OMIM phenotype	616633	117	100	96	90
PRSS12	Mental retardation 1, 249500	606709	94	100	100	99
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	121	100	100	97
PSMD12	Stankiewicz-Isidor syndrome, 617516	604450	47	98	86	70
PTCH1	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828	601309	91	100	98	96
PTCHD1	{Autism, susceptibility to 4}, 300830	300828	79	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	66	100	100	98
PTEN	Cowden syndrome 1, 158350 {Glioma susceptibility 2}, 613028 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 {Meningioma}, 607174 Prostate cancer, somatic, 176807	601728	115	85	78	76
PTF1A	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069	607194	136	100	100	94
PTGR1	No OMIM phenotype	601274	50	99	94	83
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	77	100	99	90
PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263	608625	79	100	100	100
PTRHD1	No OMIM phenotype	617342	119	100	100	100
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	86	100	100	95
PUF60	Verheij syndrome, 615583	604819	130	100	100	100
PUM1	Spinocerebellar ataxia 47, 617931	607204	67	100	100	98
PURA	Mental retardation 31, 616158	600473	206	100	100	100
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	88	100	99	95
PUS3	Mental retardation 55, 617051	616283	70	100	99	96
PUS7	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342	616261	59	100	98	90
PYCR1	Cutis laxa, type IIB, 612940 Cutis laxa, type IIIB, 614438	179035	93	100	100	100
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	114	100	100	100
QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	108	100	100	100
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	94	100	100	97
RAB18	Warburg micro syndrome 3, 614222	602207	85	100	100	97
RAB27A	Griscelli syndrome, type 2, 607624	603868	54	100	100	91
RAB39B	Mental retardation 72, 300271 Waisman syndrome, 311510	300774	59	100	100	100
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	67	100	100	97
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	66	100	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
RAB40AL	No OMIM phenotype	300405	197	100	100	100
RAC1	Mental retardation 48, 617751	602048	84	100	100	97
RAD21	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376	606462	69	100	99	92
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	98	100	100	98
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	78	100	99	95
RAI1	Smith-Magenis syndrome, 182290	607642	187	100	100	100
RARB	Microphthalmia, syndromic 12, 615524	180220	85	100	100	99
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	66	100	99	93
RBBP8	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744	604124	57	100	99	92
RBFOX1	No OMIM phenotype	605104	116	100	100	99
RBM10	TARP syndrome, 311900	300080	81	100	96	88
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	70	100	100	96
RBPJ	Adams-Oliver syndrome 3, 614814	147183	77	100	99	89
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175	607867	98	100	100	99
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	74	100	100	97
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975	605226	65	97	92	86
REV3L	No OMIM phenotype	602776	61	100	99	95
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	67	100	99	93
RHEB	No OMIM phenotype	601293	27	86	60	36
RHOBTB2	Epileptic encephalopathy, early infantile, 64, 618004	607352	151	100	100	99
RIT1	Noonan syndrome 8, 615355	609591	72	100	100	100
RLIM	Tonne-Kalscheuer syndrome, 300978	300379	68	100	100	90
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	67	100	100	93
RMRP	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460	157660	No coverage data			
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	109	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	62	100	98	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	323	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	106	100	100	99
RNF113A	?Trichothiodystrophy 5, nonphotosensitive, 300953	300951	107	100	100	100
RNF125	Tenorio syndrome, 616260	610432	94	100	100	100
RNF135	No OMIM phenotype	611358	83	100	100	99
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	90	100	98	93
RORA	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060	600825	82	100	100	97
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	60	98	96	89
RPL10	{Autism, susceptibility to 5}, 300847 Mental retardation, syndromic, 35, 300998	312173	83	100	100	100
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation 19, 300844	300075	46	100	92	70
RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723	616585	76	100	98	90
RTEL1	Dyskeratosis congenita 4, 615190 Dyskeratosis congenita 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	131	100	100	99
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833	610436	71	100	99	94
RUBCN	?Spinocerebellar ataxia 15, 615705	613516	85	100	98	96
RUSC2	Mental retardation 61, 617773	611053	168	100	100	100
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	90	100	100	94
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480	602218	120	100	100	99
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	64	100	99	88
SATB2	Glass syndrome, 612313	608148	95	100	100	97
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	79	100	100	99
SC5D	Lathosterolosis, 607330	602286	88	100	100	100
SCAF4	No OMIM phenotype	616023	65	100	98	90
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634	182389	92	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SCN1B	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350	600235	169	100	97	93
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	95	100	100	97
SCN3A	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938	182391	84	100	100	97
SCN8A	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 ?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080	600702	110	100	100	98
SCO1	Mitochondrial complex IV deficiency, 220110	603644	99	100	100	99
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	125	100	100	100
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	134	100	97	91
SEMA3E	?CHARGE syndrome, 214800	608166	64	100	100	94
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	78	100	100	96
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	65	100	99	86
SETBP1	Mental retardation 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	102	99	98	97
SETD1A	No OMIM phenotype	611052	135	100	99	98
SETD1B	No OMIM phenotype	611055	150	97	96	95
SETD2	Luscan-Lumish syndrome, 616831	612778	68	100	100	98
SETD5	Mental retardation 23, 615761	615743	85	100	100	98
SF1	No OMIM phenotype	601516	58	90	84	79
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	103	100	94	90
SH3KBP1	?Immunodeficiency 61, 300310	300374	51	99	89	69
SHANK2	{Autism susceptibility 17}, 613436	603290	123	100	99	96
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	117	99	93	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	119	100	100	99
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	61	100	99	95
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434	300579	70	100	100	97
SIK1	Epileptic encephalopathy, early infantile, 30, 616341	605705	125	100	100	99
SIL1	Marinesco-Sjogren syndrome, 248800	608005	100	100	100	100
SIN3A	Witteveen-Kolk syndrome, 613406	607776	67	100	100	97
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	158	100	100	96
SKI	Shprintzen-Goldberg syndrome, 182212	164780	123	100	100	99
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	67	100	99	94
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905	608305	126	100	100	98
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	75	100	100	95
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	86	100	100	97
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	73	100	100	98
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	93	100	100	98
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105	600300	68	100	100	98
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	600229	136	100	100	100
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	88	100	100	97
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	132	100	100	98
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	105	100	100	100
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	122	100	100	100
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	127	100	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	68	100	98	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	66	100	100	97
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896, Somatic mosaicism	314375	75	100	100	99
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	73	100	99	92
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	146	100	100	100
SLC39A12	No OMIM phenotype	608734	56	100	97	87
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721	608732	83	100	99	95
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	61	100	100	97
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	115	100	100	100
SLC6A17	Mental retardation 48, 616269	610299	119	100	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135	126455	102	100	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	103	100	98	95
SLC7A7	Lysinuric protein intolerance, 222700	603593	79	100	100	97
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	64	100	96	84
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	79	100	100	98
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	94	98	98	96
SMARCA4	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325	603254	127	100	100	100
SMARCB1	Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322 Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091	601607	130	100	100	99
SMARCC2	Coffin-Siris syndrome 8, 618362	601734	74	100	100	94
SMARCE1	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174	603111	57	100	100	93
SMC1A	Cornelia de Lange syndrome 2, 300590	300040	71	100	100	97
SMC3	Cornelia de Lange syndrome 3, 610759	606062	69	100	97	88
SMOC1	Microphthalmia with limb anomalies, 206920	608488	106	100	100	97
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	131	100	100	99
SMPD4	No OMIM phenotype	610457	112	100	100	98
SMS	Mental retardation, Snyder-Robinson type, 309583	300105	42	95	89	68

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SNAP25	?Myasthenic syndrome, congenital, 18, 616330	600322	84	100	100	98
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	148	100	100	100
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	100	100	100	100
SNRNP70	No OMIM phenotype	180740	102	100	100	98
SNRPN	Prader-Willi syndrome, 176270	182279	96	100	100	100
SNX14	Spinocerebellar ataxia 20, 616354	616105	64	100	99	91
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	165	100	98	95
SON	ZTTK syndrome, 617140	182465	85	99	96	90
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	71	100	100	95
SOS2	Noonan syndrome 9, 616559	601247	75	100	99	95
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	72	100	97	89
SOX11	Coffin-Siris syndrome 9, 615866	600898	176	100	100	100
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	183	100	100	100
SOX3	Mental retardation, with isolated growth hormone deficiency, 300123 Panhypopituitarism, 312000	313430	64	100	95	89
SOX5	Lamb-Shaffer syndrome, 616803	604975	78	100	100	96
SPAST	Spastic paraplegia 4, 182601	604277	64	100	100	94
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577	613940	68	100	100	97
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	84	100	100	97
SPOCK1	No OMIM phenotype	602264	107	100	100	97
SPRED1	Legius syndrome, 611431	609291	57	100	98	92
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	95	100	100	98
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia 14, 615386	604985	121	100	100	100
SRCAP	Floating-Harbor syndrome, 136140	611421	139	100	100	100
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	111	100	100	96
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	59	100	99	95
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	76	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation 12, 611090	606494	79	100	100	96
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	55	99	92	84
STAG1	Mental retardation 47, 617635	604358	49	100	98	83
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	72	100	100	97
STIL	Microcephaly 7, primary, 612703	181590	65	100	100	97
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	98	100	100	100
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	96	100	100	98
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	63	100	100	98
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	90	100	100	97
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172	601485	131	100	100	99
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	76	100	100	99
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	56	100	97	86
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	88	100	100	100
SUCO	No OMIM phenotype	No ID	62	100	99	94
SUMF1	Multiple sulfatase deficiency, 272200	607939	88	100	100	99
SUOX	Sulfite oxidase deficiency, 272300	606887	136	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	88	94	89	87
SUZ12	No OMIM phenotype	606245	69	100	98	92
SYN1	Epilepsy, with variable learning disabilities and behavior disorders, 300491 dominant	313440	60	100	99	85
SYNCRIP	No OMIM phenotype	616686	66	97	89	76
SYNE1	Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, 612998 Spinocerebellar ataxia 8, 610743	608441	77	100	100	97
SYNGAP1	Mental retardation 5, 612621	603384	158	98	98	98
SYP	Mental retardation 96, 300802	313475	69	100	100	98
SYT1	Baker-Gordon syndrome, 618218	185605	98	100	100	99
SYT14	?Spinocerebellar ataxia 11, 614229	610949	61	100	95	86
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	117	100	100	100
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	54	100	97	87
TAF2	Mental retardation 40, 615599	604912	64	100	99	92

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TAT	Tyrosinemia, type II, 276600	613018	65	100	100	98
TBC1D20	Warburg micro syndrome 4, 615663	611663	74	100	93	93
TBC1D24	DOORS syndrome, 220500 Deafness 86, 614617 Deafness 65, 616044 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	158	100	100	100
TBC1D7	Macrocephaly/megalencephaly syndrome, 248000	612655	55	100	98	85
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	604649	104	100	99	96
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	59	100	97	88
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	616899	53	100	99	91
TBL1XR1	Mental retardation 41, 616944 Pierpont syndrome, 602342	608628	71	100	98	90
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	80	100	100	96
TBR1	Intellectual developmental disorder with autism and speech delay, 606053	604616	148	100	100	99
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430	602054	94	94	85	80
TCF12	Craniosynostosis 3, 615314	600480	72	100	100	98
TCF20	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430	603107	107	100	100	100
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954	602272	73	100	100	96
TCF7L2	{Diabetes mellitus, type 2, susceptibility to}, 125853	602228	110	100	100	97
TCN2	Transcobalamin II deficiency, 275350	613441	118	100	100	100
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	66	100	100	97
TDP2	Spinocerebellar ataxia 23, 616949	605764	120	100	100	99
TECPR2	Spastic paraplegia 49, 615031	615000	102	100	100	99
TECR	Mental retardation 14, 614020	610057	140	100	100	100
TELO2	You-Hoover-Fong syndrome, 616954	611140	108	100	97	93

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TFAP2A	Branchiooculofacial syndrome, 113620	107580	95	100	100	100
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	171	94	93	93
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	210	100	100	100
TGIF1	Holoprosencephaly 4, 142946	602630	152	100	100	100
TH	Segawa syndrome, recessive, 605407	191290	93	100	99	94
THOC2	Mental retardation 12/35, 300957	300395	43	100	94	72
THOC6	Beaulieu-Boycott-Innes syndrome, 613680	615403	210	100	100	100
THRB	Thyroid hormone resistance, 188570 Thyroid hormone resistance, 274300 Thyroid hormone resistance, selective pituitary, 145650	190160	88	100	100	99
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	138	100	100	100
TINF2	Dyskeratosis congenita 3, 613990 Revesz syndrome, 268130	604319	155	100	100	100
TLK2	Mental retardation 57, 618050	608439	55	97	82	66
TM4SF20	{Specific language impairment 5}, 615432	615404	58	100	100	96
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614123	108	100	100	96
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	108	100	100	100
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	96	100	95	90
TMEM237	Joubert syndrome 14, 614424	614423	57	100	98	87
TMEM240	Spinocerebellar ataxia 21, 607454	616101	108	100	100	100
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 ?RHYNS syndrome, 602152	609884	77	100	100	94
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	93	100	100	95
TMLHE	{Autism, susceptibility to 6}, 300872	300777	32	89	81	67
TMPRSS7	No OMIM phenotype	No ID	51	100	99	92
TMTC3	Lissencephaly 8, 617255	617218	73	100	99	93
TMX2	No OMIM phenotype	616715	69	100	94	88

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TNIK	Mental retardation 54, 617028	610005	56	100	98	92
TOE1	Pontocerebellar hypoplasia, type 7, 614969	613931	110	100	100	98
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	123	100	98	96
TPO	Thyroid dyshormonogenesis 2A, 274500	606765	116	100	100	100
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	97	100	100	100
TRAIP	Seckel syndrome 9, 616777	605958	98	100	100	100
TRAPPC11	Muscular dystrophy, limb-girdle 18, 615356	614138	60	100	97	90
TRAPPC6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862	610397	56	100	95	78
TRAPPC9	Mental retardation 13, 613192	611966	100	100	100	98
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	234	100	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle 8, 254110	602290	107	100	100	100
TRIO	Mental retardation 44, 617061	601893	100	99	99	97
TRIP12	Mental retardation 49, 617752	604506	75	100	99	95
TRMT1	Mental retardation 68, 618302	611669	107	100	99	94
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	616013	91	100	100	94
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	167	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	184	100	100	100
TSEN15	Pontocerebellar hypoplasia, type 2F, 617026	608756	70	100	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	105	100	96	96
TSHB	Hypothyroidism, congenital, nongoitrous 4, 275100	188540	95	100	100	100
TSPAN7	Mental retardation 58, 300210	300096	55	100	98	83
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	53	100	92	77
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	58	100	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	66	100	100	93
TTI2	Mental retardation 39, 615541	614426	66	100	100	98
TUBA1A	Lissencephaly 3, 611603	602529	110	100	100	100
TUBA8	Cortical dysplasia, complex, with other brain malformations 8, 613180	605742	118	100	100	100
TUBB	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610	191130	198	100	99	97
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	99	99	82	74
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, 610031	612850	119	100	87	78
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	269	100	99	95
TUBB4A	Dystonia 4, torsion, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	242	100	100	99
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	190	100	100	100
TUBGCP4	Microcephaly and chorioretinopathy, 3, 616335	609610	71	100	98	94
TUBGCP6	Microcephaly and chorioretinopathy, 1, 251270	610053	159	100	100	99
TUSC3	Mental retardation 7, 611093	601385	72	100	100	95
TWIST1	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746	601622	132	100	100	95
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	145	100	100	100
UBA5	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia 24, 617133	610552	46	100	91	67
UBE2A	Mental retardation syndromic, Nascimento-type, 300860	312180	57	100	99	81
UBE3A	Angelman syndrome, 105830	601623	61	100	100	96
UBE3B	Kaufman oculocerebrofacial syndrome, 244450	608047	116	100	100	99
UBR1	Johanson-Blizzard syndrome, 243800	605981	61	100	99	93
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801	612636	75	100	100	97
UPB1	Beta-ureidopropionase deficiency, 613161	606673	96	100	100	100
UPF3B	Mental retardation, syndromic 14, 300676	300298	74	100	98	87
UQCC2	Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	127	100	100	100
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	123	100	100	100

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UROC1	?Urocanase deficiency, 276880	613012	112	100	100	99
USP18	Pseudo-TORCH syndrome 2, 617397	607057	117	95	95	93
USP27X	Mental retardation 105, 300984	300975	77	100	100	100
USP7	No OMIM phenotype	602519	44	97	89	71
USP9X	Mental retardation 99, 300919 Mental retardation 99, syndromic, female-restricted, 300968	300072	67	100	98	94
UTRN	No OMIM phenotype	128240	77	100	100	97
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	70	100	100	97
VPS11	Leukodystrophy, hypomyelinating, 12, 616683	608549	102	100	100	98
VPS13B	Cohen syndrome, 216550	607817	75	100	99	96
VPS13C	Parkinson disease 23, early onset, 616840	608879	50	100	97	85
VPS37A	Spastic paraplegia 53, 614898	609927	46	100	99	86
VPS53	Pontocerebellar hypoplasia, type 2E, 615851	615850	82	100	100	96
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	53	100	99	92
VWA3B	?Spinocerebellar ataxia 22, 616948	614884	66	100	98	92
WAC	Desanto-Shinawi syndrome, 616708	615049	57	100	98	91
WASHC4	?Mental retardation 43, 615817	615748	66	100	97	90
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	62	100	100	94
WBP11	No OMIM phenotype	618083	49	96	74	54
WDR13	No OMIM phenotype	300512	91	100	100	98
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	71	100	100	97
WDR26	Skraban-Deardorff syndrome, 617616	617424	50	100	94	81
WDR4	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346	605924	117	100	100	100
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	92	100	100	100
WDR54	No OMIM phenotype	No ID	105	100	100	99
WDR62	Microcephaly 2, primary, with or without cortical malformations, 604317	613583	139	100	100	100
WDR73	Galloway-Mowat syndrome 1, 251300	616144	152	100	100	97
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967	614218	155	100	100	100

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WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, 614296	606201	196	100	100	100
WNK3	No OMIM phenotype	300358	44	100	95	82
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia 12, 614322	605131	92	100	100	100
XIAP	Lymphoproliferative syndrome, 2, 300635	300079	47	100	95	80
XPA	Xeroderma pigmentosum, group A, 278700	611153	71	100	100	93
XPC	Xeroderma pigmentosum, group C, 278720	613208	108	100	100	98
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	63	100	100	96
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	103	100	97	93
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	73	100	99	94
YME1L1	?Optic atrophy 11, 617302	607472	48	100	95	83
YWHAE	No OMIM phenotype	605066	75	100	100	91
YY1	Gabriele-de Vries syndrome, 617557	600013	97	100	100	100
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	128	100	100	100
ZBTB18	Mental retardation 22, 612337	608433	157	100	100	99
ZBTB20	Primrose syndrome, 259050	606025	149	100	100	100
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069	614064	73	100	100	100
ZC3H14	Mental retardation 56, 617125	613279	93	100	99	93
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	78	100	99	93
ZDHHC15	No OMIM phenotype	300576	48	100	99	89
ZDHHC9	Mental retardation syndromic, Raymond type, 300799	300646	44	100	97	83
ZEB2	Mowat-Wilson syndrome, 235730	605802	85	100	100	100
ZFYVE26	Spastic paraplegia 15, 270700	612012	90	100	100	98
ZIC1	Craniosynostosis 6, 616602	600470	255	100	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	161	96	95	93
ZIC4	No OMIM phenotype	608948	132	100	100	99
ZMYND11	Mental retardation 30, 616083	608668	73	100	100	98
ZNF101	No OMIM phenotype	603983	59	100	100	93

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ZNF292	No OMIM phenotype	616213	58	100	98	95
ZNF407	No OMIM phenotype	615894	86	100	100	100
ZNF41	No OMIM phenotype	314995	49	100	99	93
ZNF592	No OMIM phenotype	613624	141	100	100	100
ZNF674	No OMIM phenotype	300573	57	100	99	93
ZNF711	Mental retardation 97, 300803	314990	54	100	96	85
ZNF81	No OMIM phenotype	314998	49	100	100	90
ZSWIM6	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865	615951	71	96	93	88

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x