Whole Exome Sequencing
Gene package Intellectual disability, version 5.1, 22-11-2017

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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).

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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	No OMIM phenotype	610627	44	100	97	80
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	59	100	97	86
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	45	100	95	80
ABAT	GABA-transaminase deficiency, 613163	137150	65	100	98	87
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	46	100	95	80
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	56	83	72	70
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	71	100	100	94
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	53	100	99	89
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	64	100	100	98
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	93	100	100	100
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	71	100	100	94
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	86	100	100	96
ACSL4	Mental retardation 63, 300387	300157	51	100	98	87

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ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	135	100	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	121	100	100	100
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	51	100	99	89
ACY1	Aminoacylase 1 deficiency, 609924	104620	72	100	100	100
ADA2	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	62	100	99	87
ADAM22	No OMIM phenotype	603709	46	100	97	78
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	67	100	100	98
ADAT3	Mental retardation 36, 615286	615302	84	100	100	100
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	67	100	100	98
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	38	100	90	62
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	61	100	100	99
ADSL	Adenylosuccinase deficiency, 103050	608222	61	100	99	88
AFF2	Mental retardation, FRAXE type, 309548	300806	46	100	96	84
AFF4	CHOPS syndrome, 616368	604417	54	100	97	85
AFG3L2	Spastic ataxia 5, 614487 Spinocerebellar ataxia 28, 610246	604581	54	99	93	79
AGA	Aspartylglucosaminuria, 208400	613228	53	100	98	85
AGAP2	No OMIM phenotype	605476	79	100	99	96
AGO2	No OMIM phenotype	606229	56	99	92	78
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	57	100	100	96
AGTR2	No OMIM phenotype	300034	53	100	100	99
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	93	96	96	95
AHDC1	Xia-Gibbs syndrome, 615829	615790	113	100	99	98
AHI1	Joubert syndrome 3, 608629	608894	51	100	94	73
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness 5, 300614	300169	41	100	89	67
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	62	100	100	94
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	80	100	100	100

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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	72	100	100	97
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	49	100	94	75
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	54	100	98	88
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	52	100	98	88
ALDH4A1	Hyperprolinemia, type II, 239510	606811	64	100	99	97
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	42	99	94	71
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	46	90	76	70
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	59	100	100	99
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	108	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	42	100	96	73
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	51	100	100	97
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	63	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	58	100	99	83
ALG8	Congenital disorder of glycosylation, type Ih, 608104	608103	43	100	96	75
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	43	100	96	77
ALMS1	Alstrom syndrome, 203800	606844	82	100	99	96
ALX1	?Frontonasal dysplasia 3, 613456	601527	57	100	100	99
ALX4	{Craniosynostosis 5, susceptibility to}, 615529 Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597	605420	83	100	100	100
AMMECR1	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990	300195	41	97	79	67
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686	102771	80	100	100	99
AMT	Glycine encephalopathy, 605899	238310	73	100	100	98
ANK3	?Mental retardation, 37, 615493	600465	56	100	99	94
ANKEF1	No OMIM phenotype	No id	61	100	99	93

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ANKH	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	605145	59	100	100	98
ANKLE2	?Microcephaly 16, primary, 616681	616062	70	99	93	87
ANKRD11	KBG syndrome, 148050	611192	89	100	100	100
ANO10	Spinocerebellar ataxia 10, 613728	613726	40	100	92	65
ANTXR1	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089	606410	38	98	92	68
AP1S1	MEDNIK syndrome, 609313	603531	51	100	100	95
AP1S2	Mental retardation syndromic 5, 304340	300629	44	100	80	65
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	50	100	95	71
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276	602166	72	100	99	95
AP4B1	Spastic paraplegia 47, 614066	607245	60	100	100	96
AP4E1	Spastic paraplegia 51, 613744 Stuttering, familial persistent, 1, 184450	607244	50	100	99	87
AP4M1	Spastic paraplegia 50, 612936	602296	75	100	100	98
AP4S1	Spastic paraplegia 52, 614067	607243	37	100	95	69
APC2	?Sotos syndrome 3, 617169	612034	68	98	94	89
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	54	100	90	77
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	55	100	98	85
ARG1	Argininemia, 207800	608313	56	100	100	88
ARHGAP4	No OMIM phenotype	300023	56	100	99	92
ARHGEF6	Mental retardation 46, 300436	300267	38	100	93	70
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	38	100	95	63
ARID1A	Coffin-Siris syndrome 2, 614607	603024	75	100	98	94
ARID1B	Coffin-Siris syndrome 1, 135900	614556	67	99	97	90
ARID2	No OMIM phenotype	609539	59	100	99	91
ARL13B	Joubert syndrome 8, 612291	608922	50	100	99	86
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	32	97	88	57
ARNT2	?Webb-Dattani syndrome, 615926	606036	55	100	98	88
ARSA	Metachromatic leukodystrophy, 250100	607574	105	100	100	100
ARSE	Chondrodysplasia punctata recessive, 302950	300180	63	100	95	84

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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	42	89	80	65
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	49	100	97	80
ASCL1	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880	100790	120	100	100	100
ASL	Argininosuccinic aciduria, 207900	608310	70	100	100	99
ASNS	Asparagine synthetase deficiency, 615574	108370	59	100	100	92
ASPA	Canavan disease, 271900	608034	41	100	98	81
ASPM	Microcephaly 5, primary, 608716	605481	62	100	100	96
ASS1	Citrullinemia, 215700	603470	70	100	100	98
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	77	100	100	97
ASXL2	Shashi-Pena syndrome, 617190	612991	69	100	99	90
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	52	99	98	94
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	68	100	96	92
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	45	100	99	84
ATN1	Dentatorubro-pallidoluysian atrophy, 125370	607462	80	100	100	97
ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	86	100	100	99
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	91	100	100	100
ATP2A2	Acrokeratosis verruciformis, 101900 Darier disease, 124200	108740	67	100	100	98
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	45	100	93	64
ATP6V0A2	Cutis laxa, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	56	100	98	87
ATP6V1B2	Deafness, congenital, with onychodystrophy, 124480 Zimmermann-Laband syndrome 2, 616455	606939	59	100	100	97

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ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	44	100	98	84
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	48	100	98	81
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	51	100	100	90
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600	601215	58	100	96	78
ATRIP	No OMIM phenotype	606605	73	100	98	92
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	40	100	91	76
AUH	3-methylglutaconic aciduria, type I, 250950	600529	52	100	100	92
AUTS2	Mental retardation 26, 615834	607270	60	100	97	91
AVPR2	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	300538	84	100	100	95
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	42	100	97	76
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	48	79	76	72
B3GLCT	Peters-plus syndrome, 261540	610308	47	100	87	64
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	69	100	100	94
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070	604327	76	100	100	97
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	114	100	100	100
BBS1	Bardet-Biedl syndrome 1, 209900	209901	69	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	55	100	100	98
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	96
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	58	100	99	87
BBS4	Bardet-Biedl syndrome 4, 615982	600374	53	100	96	81
BBS5	Bardet-Biedl syndrome 5, 615983	603650	51	100	99	82
BBS7	Bardet-Biedl syndrome 7, 615984	607590	51	100	98	87
BBS9	Bardet-Biedl syndrome 9, 615986	607968	42	96	94	79
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	48	100	99	83
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	86	100	100	100
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	49	100	99	86
BCL11A	Dias-Logan syndrome, 617101	606557	89	100	100	99
BCOR	Microphthalmia, syndromic 2, 300166	300485	77	100	97	92
BCORL1	No OMIM phenotype	300688	74	100	99	94

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BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	134	100	100	100
BLM	Bloom syndrome, 210900	604610	62	100	98	90
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	53	100	98	84
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	74	100	100	98
BRF1	Cerebellofaciodental syndrome, 616202	604902	68	100	99	91
BRPF1	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333	602410	74	100	100	98
BRWD3	Mental retardation 93, 300659	300553	41	100	97	78
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	68	100	100	99
BTD	Biotinidase deficiency, 253260	609019	70	100	100	98
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	51	100	98	86
c12orf4	No OMIM phenotype	616082	57	100	99	84
C12orf57	Temtamy syndrome, 218340	615140	103	100	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, 615035	613541	57	100	100	100
C1orf167	No OMIM phenotype	No id	66	100	98	92
C2CD3	?Orofaciodigital syndrome XIV, 615948	615944	58	100	99	92
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	54	100	98	88
CA2	Osteopetrosis 3, with renal tubular acidosis, 259730	611492	57	100	100	96
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	110	100	100	100
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	51	100	95	81

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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	63	100	98	90
CACNA1C	Brugada syndrome 3, 611875 Timothy syndrome, 601005	114205	66	100	99	95
CACNA2D1	No OMIM phenotype	114204	43	100	96	75
CACNG2	?Mental retardation 10, 614256	602911	78	100	99	94
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	76	100	100	100
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	90	100	98	94
CAPN10	{Diabetes mellitus, noninsulin-dependent 1}, 601283	605286	81	100	100	100
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	42	100	95	69
CBL	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	54	100	100	97
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	70	100	100	96
CC2D1A	Mental retardation 3, 608443	610055	79	100	100	99
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	48	100	97	81
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	58	100	98	90
CCDC13	No OMIM phenotype	No id	52	100	100	93
CCDC14	No OMIM phenotype	617147	55	100	100	92
CCDC22	Ritscher-Schinzel syndrome 2, 300963	300859	58	100	97	88
CCDC78	Myopathy, centronuclear, 4, 614807	614666	80	100	100	100
CCDC88C	Hydrocephalus, nonsyndromic, 236600 ?Spinocerebellar ataxia 40, 616053	611204	67	100	99	94
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	123833	68	100	100	98
CDC5L	No OMIM phenotype	602868	39	100	96	75
CDH15	Mental retardation 3, 612580	114019	67	100	99	94
CDK16	No OMIM phenotype	311550	63	100	100	98
CDK5	?Lissencephaly 7 with cerebellar hypoplasia, 616342	123831	78	100	100	100
CDK5RAP2	Microcephaly 3, primary, 604804	608201	47	100	95	78
CDK6	?Microcephaly 12, primary, 616080	603368	49	100	98	90
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	46	100	93	78

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CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	56	91	83	74
CDON	Holoprosencephaly 11, 614226	608707	62	100	100	92
CENPJ	Microcephaly 6, primary, 608393 ?Seckel syndrome 4, 613676	609279	60	100	100	96
CEP104	Joubert syndrome 25, 616781	616690	45	100	93	77
CEP135	Microcephaly 8, primary, 614673	611423	61	100	96	80
CEP152	Microcephaly 9, primary, 614852 Seckel syndrome 5, 613823	613529	52	100	97	84
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	57	100	98	84
CEP41	Joubert syndrome 15, 614464	610523	53	100	99	89
CEP63	?Seckel syndrome 6, 614728	614724	54	100	95	78
CEP89	No OMIM phenotype	615470	46	100	93	74
CHAMP1	Mental retardation 40, 616579	616327	85	100	100	100
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	50	100	98	85
CHD3	No OMIM phenotype	602120	57	98	93	85
CHD4	Sifrim-Hitz-Weiss syndrome, 617159	603277	50	100	99	91
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	60	100	99	93
CHD8	{Autism, susceptibility to, 18}, 615032	610528	64	100	99	92
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	65	100	100	100
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	56	100	100	96
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	108	100	95	95
CIC	No OMIM phenotype	612082	80	100	99	97
CIT	Microcephaly 17, primary, 617090	605629	61	100	97	85
CKAP2L	Filippi syndrome, 272440	616174	64	100	100	97
CLCN4	Mental retardation 49/15, 300114	302910	64	100	100	94
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	95	100	100	100
CLIC2	?Mental retardation, syndromic 32, 300886	300138	35	99	89	57
CLIP1	No OMIM phenotype	179838	58	100	95	84
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	76	100	100	94
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	57	100	100	98

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CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	78	100	98	88
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	81	100	100	100
CLP1	Pontocerebellar hypoplasia, type 10, 615803	608757	70	100	100	100
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	71	100	100	96
CNKSR2	No OMIM phenotype	300724	49	100	96	81
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	106	100	100	95
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	48	100	99	89
COASY	Neurodegeneration with brain iron accumulation 6, 615643	609855	94	100	100	100
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	66	100	99	94
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	53	100	100	89
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	43	100	96	77
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	48	100	96	74
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	56	100	98	85
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	80	100	100	99
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	47	100	96	79
COL18A1	Knobloch syndrome, type 1, 267750	120328	76	100	100	95
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000	120130	61	100	99	91
COL4A2	{Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 2, 614483	120090	67	100	100	96
COL4A3BP	Mental retardation 34, 616351	604677	44	100	92	74
COLEC11	3MC syndrome 2, 265050	612502	84	100	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	49	100	97	82
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276	612898	74	100	100	100
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	86	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
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	71	100	100	95
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	93	100	100	95
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	43	100	96	76
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	60	100	100	100
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	42	100	96	78
CRADD	Mental retardation 34, with variant lissencephaly, 614499	603454	96	100	100	100
CRB2	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	609720	76	100	100	99
CRBN	Mental retardation 2, 607417	609262	50	100	96	76
CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	81	100	99	91
CRLF1	Cold-induced sweating syndrome 1, 272430	604237	72	94	90	87
CSNK2A1	Okur-Chung neurodevelopmental syndrome, 617062	115440	45	100	90	65
CSPP1	Joubert syndrome 21, 615636	611654	58	100	99	89
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	62	100	100	98
CTBP1	No OMIM phenotype	602618	50	96	92	75
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	72	100	100	98
CTCF	Mental retardation 21, 615502	604167	70	100	100	99
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	78	100	91	86
CTNNB1	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	47	100	100	92
CTNND1	No OMIM phenotype	601045	52	100	99	88
CTNND2	No OMIM phenotype	604275	66	99	95	87
CTSA	Galactosialidosis, 256540	613111	83	100	100	98
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	79	100	100	100
CTTNBP2	No OMIM phenotype	609772	72	100	99	91
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	59	100	98	84
CUL4B	Mental retardation, syndromic 15 (Cabezas type), 300354	300304	49	100	93	77
CWF19L1	Spinocerebellar ataxia 17, 616127	616120	40	100	94	71
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	76	100	100	99

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CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	82	100	100	100
CYP2U1	Spastic paraplegia 56, 615030	610670	51	100	100	91
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	82	100	100	100
DAB1	No OMIM phenotype	603448	59	100	100	93
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	128239	103	100	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	45	100	98	81
DBT	Maple syrup urine disease, type II, 248600	248610	77	100	100	97
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	56	100	96	83
DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Mirror movements 1, 157600	120470	48	100	99	89
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	94	100	100	100
DCPS	Al-Raqad syndrome, 616459	610534	71	100	100	100
DCX	Lissencephaly, 300067 Subcortical laminal heteropia, 300067	300121	47	100	96	83
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	55	100	93	82
DDHD2	Spastic paraplegia 54, 615033	615003	57	100	100	93
DDX11	Warsaw breakage syndrome, 613398	601150	159	100	100	100
DDX3X	Mental retardation 102, 300958 dominant	300160	76	100	99	97
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation 24, 615828	602635	56	99	85	84
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281	617278	44	100	95	75
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	56	100	98	90
DHCR24	Desmosterolosis, 602398	606418	71	100	100	99
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	84	100	100	100
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	65	100	100	96
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	54	100	99	91
DIAPH1	Deafness 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	60	100	95	83
DIP2B	Mental retardation, FRA12A type, 136630	611379	46	100	97	81
DKC1	Dyskeratosis congenita, 305000	300126	45	100	96	83
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	61	100	100	94
DLG3	Mental retardation 90, 300850	300189	51	100	95	83
DLG4	No OMIM phenotype	602887	89	100	100	99

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DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	38	100	94	71
DMPK	Myotonic dystrophy 1, 160900	605377	69	100	99	93
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	50	100	94	76
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	56	100	100	82
DNM1	Epileptic encephalopathy, early infantile, 31, 616346	602377	82	100	99	92
DNMT3A	Tatton-Brown-Rahman syndrome, 615879	602769	67	100	100	96
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	71	100	99	92
DOCK6	Adams-Oliver syndrome 2, 614219	614194	72	100	99	98
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859	615730	43	100	96	77
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	49	100	94	81
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	94	100	100	100
DONSON	No OMIM phenotype	611428	35	100	91	63
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	54	100	100	95
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901	603527	73	100	100	100
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	54	93	89	77
DPP6	Mental retardation 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956	126141	49	100	92	78
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	46	100	99	85
DPYS	Dihydropyrimidinuria, 222748	613326	55	100	99	90
DST	Epidermolysis bullosa simplex 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653	113810	51	100	98	86
DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	49	100	93	68
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	69	100	99	95
DYRK1A	Mental retardation 7, 614104	600855	54	100	98	87
EBP	Chondrodysplasia punctata dominant, 302960 MEND syndrome, 300960	300205	66	100	100	96
EDC3	?Mental retardation 50, 616460	609842	79	100	100	99
EDNRB	ABCD syndrome, 600501 {Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580	131244	69	100	100	99
EDRF1	No OMIM phenotype	No id	48	100	99	87

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EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation 38, 616393	602959	93	100	100	98
EFCAB1	No OMIM phenotype	No id	47	100	100	85
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536	603892	58	100	99	90
EHMT1	Kleefstra syndrome, 610253	607001	81	99	99	98
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	53	100	95	83
EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305	608546	44	100	98	76
EIF4G1	{Parkinson disease 18}, 614251	600495	73	100	100	99
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731	605367	55	100	99	92
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	56	100	100	95
ELP2	Mental retardation 58, 617270	616054	55	100	100	95
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	80	100	99	92
EMG1	Bowen-Conradi syndrome, 211180	611531	57	100	100	100
EML1	Band heterotopia, 600348	602033	50	100	97	85
EMX2	Schizencephaly, 269160	600035	83	100	100	98
ENTPD1	Spastic paraplegia 64, 615683	601752	46	100	97	79
EOMES	No OMIM phenotype	604615	63	100	100	98
EP300	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684	602700	66	100	98	90
EPB41L1	?Mental retardation 11, 614257	602879	74	100	99	98
EPG5	Vici syndrome, 242840	615068	49	100	97	84
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	52	100	100	95
ERCC2	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	75	100	100	99
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	133510	56	100	99	89
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	133530	65	100	99	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	60	100	98	92
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	55	100	93	79
ERLIN2	Spastic paraplegia 18, 611225	611605	47	100	99	84
ERMARD	?Periventricular nodular heterotopia 6, 615544	615532	51	100	98	87
ESCO2	Roberts syndrome, 268300 SC phocomelia syndrome, 269000	609353	48	100	99	87
ETFB	Glutaric acidemia IIB, 231680	130410	68	100	100	100
ETHE1	Ethylmalonic encephalopathy, 602473	608451	71	100	100	92
EXOSC2	No OMIM phenotype	602238	49	100	99	86
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	59	100	100	87
EZH2	Weaver syndrome, 277590	601573	56	100	99	88
FA2H	Spastic paraplegia 35, 612319	611026	56	100	100	92
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	50	100	100	94
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154	616107	58	100	100	92
FAT2	No OMIM phenotype	604269	73	100	100	97
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	69	100	100	98
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	111	100	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	51	100	98	89
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	605654	51	100	100	92
FBXO31	?Mental retardation 45, 615979	609102	64	100	99	92
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation syndromic 16, 305400	300546	60	100	99	95

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FGF12	Epileptic encephalopathy, early infantile, 47, 617166	601513	36	100	90	56
FGF14	Spinocerebellar ataxia 27, 609307	601515	47	100	98	83
FGFR1	Encephalocraniocutaneous lipomatosis, 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	70	100	100	96
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	49	100	98	81
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	84	100	100	98
FGL1	No OMIM phenotype	605776	44	100	99	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	57	98	93	88
FIBP	Thauvin-Robinet-Faivre syndrome, 617107	608296	67	100	100	98
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	40	100	96	77
FIGN	No OMIM phenotype	605295	74	100	100	99
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	99	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	65	100	100	97
FLNA	Cardiac valvular dysplasia, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	79	100	100	100
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	69	100	99	89
FMN2	Mental retardation 47, 616193	606373	56	91	87	81
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	38	100	94	68
FOXG1	Rett syndrome, congenital variant, 613454	164874	60	99	91	82
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	56	100	99	89
FOXP2	Speech-language disorder-1, 602081	605317	50	100	98	84
FRAS1	Fraser syndrome, 219000	607830	56	100	98	90
FREM2	Fraser syndrome, 219000	608945	74	100	100	98
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	616305	55	100	95	82
FRMPD4	Mental retardation 104, 300983	300838	60	100	97	87
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981	604574	33	80	64	56

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FTCD	Glutamate formiminotransferase deficiency, 229100	606806	54	97	93	89
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	59	100	100	90
FTSJ1	Mental retardation 9/44, 309549	300499	68	100	100	94
FUCA1	Fucosidosis, 230000	612280	54	100	100	87
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	52	100	100	90
GABRB1	Epileptic encephalopathy, early infantile, 45, 617153	137190	55	100	100	94
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	64	100	99	95
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	60	100	100	90
GALE	Galactose epimerase deficiency, 230350	606953	69	100	100	100
GALT	Galactosemia, 230400	606999	96	100	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	60	100	97	90
GAS6	No OMIM phenotype	600441	52	100	91	83
GATAD2B	Mental retardation 18, 615074	614998	50	100	100	91
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	44	100	100	89
GCDH	Glutaricaciduria, type I, 231670	608801	79	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	45	100	99	87
GCSH	Glycine encephalopathy, 605899	238330	70	100	91	57
GDI1	Mental retardation 41, 300849	300104	75	100	100	98
GFAP	Alexander disease, 203450	137780	54	100	100	97
GFM2	No OMIM phenotype	606544	47	100	96	78
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	79	100	100	100
GJB1	Charcot-Marie-Tooth neuropathy dominant, 1, 302800	304040	72	100	100	98
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, 613206	608803	52	97	86	75
GK	Glycerol kinase deficiency, 307030	300474	47	100	91	74

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GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	67	100	99	87
GLDC	Glycine encephalopathy, 605899	238300	50	100	94	82
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	81	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	92	100	100	97
GLYCTK	D-glyceric aciduria, 220120	610516	108	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	72	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	69	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	105	100	100	100
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	55	100	98	84
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	600998	68	100	100	96
GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 Acromegaly, somatic, 102200 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463	139320	86	100	99	97
GNB1	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation 42, 616973	139380	56	100	95	93
GNB5	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182	604447	45	100	95	77
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	59	100	98	90
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	47	100	97	86
GNS	Mucopolysaccharidosis type IIID, 252940	607664	46	100	98	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	42	100	94	79
GPHN	Molybdenum cofactor deficiency C, 615501	603930	48	100	99	84
GPT2	Mental retardation 49, 616281	138210	63	100	100	97
GRIA3	Mental retardation 94, 300699	305915	42	100	97	80
GRID2	Spinocerebellar ataxia 18, 616204	602368	55	100	98	91
GRIK2	Mental retardation, 6, 611092	138244	63	100	99	92
GRIN1	Mental retardation 8, 614254	138249	75	100	100	97
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	76	100	100	99
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	89	100	100	96
GRIN3B	No OMIM phenotype	606651	73	91	88	79
GRIP1	Fraser syndrome, 219000	604597	60	100	99	91
GRM1	Spinocerebellar ataxia 13, 614831	604473	83	100	100	95
GSE1	No OMIM phenotype	616886	61	100	99	92
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	58	100	100	95
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	41	100	100	85
GTPBP3	Combined oxidative phosphorylation deficiency 23, 616198	608536	85	100	100	99
GUCY2F	No OMIM phenotype	300041	37	100	94	72
GUSB	Mucopolysaccharidosis VII, 253220	611499	74	100	95	85
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data			
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures, 616756	610876	57	100	99	84
HAX1	Neutropenia, severe congenital 3, 610738	605998	79	100	100	99
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	35	100	90	54
HCFC1	Mental retardation 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	79	100	100	97
HCN1	Epileptic encephalopathy, early infantile, 24, 615871	602780	80	100	99	93
HDAC4	No OMIM phenotype	605314	70	100	100	98
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	300272	68	100	99	94
HDAC8	Cornelia de Lange syndrome 5, 300882	300269	33	100	95	65
HECTD1	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	617245	42	100	94	80
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	617245	48	100	92	76
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	62	100	100	90
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011	605109	45	100	97	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HERC2	Mental retardation 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	73	100	99	95
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230	601802	60	100	100	83
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	54	100	99	87
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	69	100	100	94
HIVEP2	Mental retardation 43, 616977	143054	82	100	99	96
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	63	100	99	91
HMGCL	HMG-CoA lyase deficiency, 246450	613898	57	100	100	94
HNMT	{Asthma, susceptibility to}, 600807 Mental retardation 51, 616739	605238	56	100	100	89
HNRNPH2	Mental retardation, syndromic, Bain type, 300986	300610	75	100	100	100
HNRNPK	Au-Kline syndrome, 616580	600712	35	95	77	52
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391	602869	51	100	99	84
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	104	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	72	100	100	91
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	39	100	95	75
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	105	100	100	100
HSD17B10	HDS10 mitochondrial disease, 300438	300256	63	100	100	95
HSPA9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854	600548	52	100	99	89
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, 605280	118190	56	100	100	88
HUWE1	Mental retardation syndromic, Turner type, 300706	300697	45	100	93	76
IARS	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093	600709	36	100	93	66

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
IBA57	?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, 616451	615316	89	100	100	98
IDS	Mucopolysaccharidosis II, 309900	300823	63	100	96	85
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	72	100	98	88
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	43	100	86	75
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	65	100	99	90
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	52	100	98	82
IFT81	No OMIM phenotype	605489	36	100	91	64
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	59	100	99	84
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	44	100	100	96
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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	62	100	100	92
IL1RAPL1	Mental retardation 21/34, 300143	300206	50	100	99	83
IMPA1	Mental retardation 59, 617323	602064	37	100	87	51
INPP5B	No OMIM phenotype	147264	57	100	99	89
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	80	100	100	96
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404	607875	48	100	96	85
INTS1	No OMIM phenotype	611345	64	99	97	91
INTS8	No OMIM phenotype	611351	38	100	93	62
IQSEC2	Mental retardation 1/78, 309530	300522	56	99	97	88
ISG15	Immunodeficiency 38, 616126	147571	107	100	100	100
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	58	100	99	84
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	600536	68	100	98	94
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	60	100	99	92
ITPR2	?Anhidrosis, isolated, with normal sweat glands, 106190	600144	41	100	90	67
IVD	Isovaleric acidemia, 243500	607036	86	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
JAG1	Alagille syndrome 1, 118450 ?Deafness, congenital heart defects, and posterior embryotoxon Tetralogy of Fallot, 187500	601920	62	100	100	92
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	49	100	97	83
JMJD1C	No OMIM phenotype	604503	53	100	98	89
KALRN	{Coronary heart disease, susceptibility to, 5}, 608901	604605	57	100	97	86
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900	607704	73	100	100	95
KANSL1	Koolen-De Vries syndrome, 610443	612452	74	100	100	92
KAT6A	Mental retardation 32, 616268	601408	70	100	99	95
KAT6B	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736	605880	73	100	98	92
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	80	100	100	100
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366	176262	87	100	100	100
KCNA4	No OMIM phenotype	176266	64	100	100	100
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056	600397	92	100	100	100
KCNC3	Spinocerebellar ataxia 13, 605259	176264	62	87	64	57
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500	603305	76	100	99	96
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	133	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	134	100	100	100
KCNJ6	Keppen-Lubinsky syndrome, 614098	600877	69	100	100	98
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292	605874	80	100	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	58	100	98	87
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data			
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200	602235	81	100	100	97
KCNQ3	Seizures, benign neonatal, type 2, 121201	602232	72	100	98	90
KCNQ5	No OMIM phenotype	607357	58	100	99	90
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959	608167	63	100	98	95
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	102	100	100	99
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features, 616728	609132	45	100	97	77

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
KDM4B	No OMIM phenotype	609765	62	100	97	92
KDM5C	Mental retardation, syndromic, Claes-Jensen type, 300534	314690	66	100	99	94
KDM6A	Kabuki syndrome 2, 300867	300128	48	100	95	83
KDSR	Lymphoma/leukemia, B-cell, variant	136440	47	100	99	80
KIAA1109	No OMIM phenotype	611565	57	100	99	91
KIAA1586	No OMIM phenotype	No id	58	100	100	99
KIDINS220	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296	615759	46	100	95	79
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	58	100	99	87
KIF1A	Mental retardation 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, 610357	601255	70	100	100	99
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	66	100	99	91
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	59	100	94	75
KIF4A	?Mental retardation 100, 300923	300521	43	100	95	70
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282	604593	45	100	95	76
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	66	99	95	89
KIRREL3	Mental retardation 4, 612581	607761	73	100	100	99
KLHL15	Mental retardation 103, 300982	300980	52	100	97	88
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	50	100	99	92
KMT2B	Dystonia 28, childhood-onset, 617284	606834	80	98	94	92
KMT2C	No OMIM phenotype	606833	64	100	98	90
KMT2D	Kabuki syndrome 1, 147920	602113	97	100	100	99
KNL1	Microcephaly 4, primary, 604321	609173	51	100	98	91
KPTN	Mental retardation 41, 615637	615620	73	100	100	94
KRAS	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 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	190070	61	100	97	64

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
KRBOX4	No OMIM phenotype	300585	66	100	100	96
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	78	100	100	99
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	47	100	99	89
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	57	100	98	89
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855	156225	50	100	98	86
LAMC1	No OMIM phenotype	150290	54	100	98	88
LAMC3	Cortical malformations, occipital, 614115	604349	67	100	100	98
LAMP2	Danon disease, 300257	309060	33	100	92	58
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	63	100	100	94
LARP7	Alazami syndrome, 615071	612026	50	100	98	85
LAS1L	Wilson-Turner syndrome, 309585	300964	53	100	98	81
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462	607031	56	100	99	88
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	62	100	100	100
LINS1	Mental retardation 27, 614340	610350	48	100	99	89
LMAN2L	?Mental retardation, 52, 616887	609552	49	100	97	88
LONP1	CODAS syndrome, 600373	605490	74	100	100	97
LRP2	Donnai-Barrow syndrome, 222448	600073	47	100	97	84
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	44	100	95	74
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	57	100	98	83
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	118	100	100	100
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	52	85	80	75
MAGEC3	No OMIM phenotype	300469	60	100	93	85
MAGEL2	Schaaf-Yang syndrome, 615547	605283	107	100	100	100
MAGT1	Immunodeficiency, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	42	100	99	79
MAN1B1	Mental retardation 15, 614202	604346	90	100	100	98
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	75	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	59	100	98	87
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	41	100	96	80

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	55	100	100	91
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	69	100	92	86
MAPRE2	Symmetric circumferential skin creases, congenital, 2, 616734	605789	51	100	93	68
MASP1	3MC syndrome 1, 257920	600521	70	100	100	92
MAT1A	Hypermethioninemia, persistent, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, 250850	610550	65	100	100	99
MBD5	Mental retardation 1, 156200	611472	58	100	100	97
MBOAT7	Mental retardation 57, 617188	606048	75	100	100	100
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, 308800 ?Olmsted syndrome, 300918	300294	42	100	98	81
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	48	100	99	83
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	46	100	99	86
MCM3AP	No OMIM phenotype	603294	54	100	96	86
MCOLN1	Mucolipidosis IV, 252650	605248	84	100	100	100
MCOLN3	No OMIM phenotype	607400	35	100	91	64
MCPH1	Microcephaly 1, primary, 251200	607117	59	94	94	85
MDH2	Epileptic encephalopathy, early infantile, 51, 617339	154100	59	100	100	93
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	88	100	100	94
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	57	100	100	98
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, 300895 Opitz-Kaveggia syndrome, 305450	300188	57	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	56	100	97	87
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	69	100	100	94
MED23	Mental retardation 18, 614249	605042	47	100	98	84
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589	610197	75	100	100	100
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4) Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	67	100	99	96
MEIS2	Cleft palate, cardiac defects, and mental retardation, 600987	601740	65	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
METTL23	Mental retardation 44, 615942	615262	54	100	100	96
MFSD2A	Microcephaly 15, primary, 616486	614397	69	100	100	94
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	50	100	98	81
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	80	100	100	100
MICU1	Myopathy with extrapyramidal signs, 615673	605084	36	100	94	58
MID1	Opitz GBBB syndrome, type I, 300000	300552	70	100	98	85
MID2	?Mental retardation 101, 300928	300204	45	100	93	70
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	59	100	100	99
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	81	100	100	97
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	49	100	98	86
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	56	100	97	87
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	59	100	100	97
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	76	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	51	100	98	78
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	69	100	100	98
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	50	100	100	90
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	88	100	100	99
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	60	100	100	97
MPDZ	Hydrocephalus, nonsyndromic 2, 615219	603785	49	100	97	85
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	48	100	100	97
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582	607118	44	100	97	78
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	61	100	99	84
MSL2	No OMIM phenotype	614802	66	100	100	97
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947	611766	52	100	96	72
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	65	100	100	98
MTMR9	No OMIM phenotype	606260	64	100	98	89
MTOR	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638	601231	65	100	98	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	52	100	98	85
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	61	100	98	92
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	56	100	98	86
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	58	100	100	91
MYCN	Feingold syndrome 1, 164280	164840	79	100	97	93
MYH9	Deafness 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249	160775	66	100	98	94
MYO5A	Griscelli syndrome, type 1, 214450	160777	51	100	97	84
MYT1L	Mental retardation 39, 616521	613084	67	100	99	94
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	56	100	95	92
NAA15	No OMIM phenotype	608000	59	100	98	86
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393	610672	83	100	100	99
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	72	100	100	100
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	80	100	97	90
NALCN	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	611549	50	100	99	88
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	53	100	99	88
NARS2	Combined oxidative phosphorylation deficiency 24, 616239	612803	38	100	93	67
NAT10	No OMIM phenotype	609221	54	100	97	89
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	52	100	95	76
NCOR2	No OMIM phenotype	600848	63	100	97	89
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	61	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
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	57	100	100	100
NDST1	Mental retardation 46, 616116	600853	86	100	100	99
NDUFA1	Mitochondrial complex I deficiency, 252010 dominant	300078	83	100	100	100
NDUFA11	Mitochondrial complex I deficiency, 252010 dominant	612638	72	100	100	100
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000	614530	48	100	100	79
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	87	100	100	100
NDUFAF3	Mitochondrial complex I deficiency, 252010 dominant	612911	79	100	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010 dominant	612360	54	100	100	89
NDUFS1	Mitochondrial complex I deficiency, 252010 dominant	157655	49	100	98	77
NDUFS2	Mitochondrial complex I deficiency, 252010 dominant	602985	59	100	100	97
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	603846	73	100	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	602694	56	100	100	90
NDUFS6	Mitochondrial complex I deficiency, 252010 dominant	603848	52	100	100	96
NDUFS7	Leigh syndrome, 256000	601825	64	100	100	97
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	91	100	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010 dominant	161015	95	100	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010 dominant	600532	47	100	96	78
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	54	100	100	99
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	600644	74	100	100	100
NEDD4L	Periventricular nodular heterotopia 7, 617201	606384	50	100	98	85
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	99	100	100	100
NEXMIF	Mental retardation 98, 300912	300524	51	100	100	98
NF1	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	41	98	87	67
NFATC1	No OMIM phenotype	600489	74	100	100	96
NFIA	Brain malformations and urinary tract defects, 613735	600727	55	100	98	89
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	78	100	99	90
NGLY1	Congenital disorder of deglycosylation, 615273	610661	55	100	100	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	44	100	92	84
NID1	No OMIM phenotype	131390	74	100	100	99
NIN	?Seckel syndrome 7, 614851	608684	56	100	98	84
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	50	100	96	80
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550	600635	54	100	100	96
NLGN3	{Asperger syndrome susceptibility 1}, 300494 {Autism susceptibility 1}, 300425	300336	66	100	99	94
NLGN4X	{Asperger syndrome susceptibility 2}, 300497 {Autism susceptibility 2}, 300495 Mental retardation, 300495	300427	153	100	100	100
NLRP3	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900	606416	85	100	100	100
NONO	Mental retardation, syndromic 34, 300967	300084	45	100	90	76
NOVA2	No OMIM phenotype	601991	80	97	88	85
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	58	100	100	93
NPC2	Niemann-pick disease, type C2, 607625	601015	64	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	39	100	92	67
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118	600928	54	100	97	89
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	82	100	98	91
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	42	100	100	91
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	67	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
NSD1	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	56	100	99	92
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	55	100	96	80
NSUN2	Mental retardation 5, 611091	610916	58	100	94	83
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	70	100	100	98
NUP62	Striatonigral degeneration, infantile, 271930	605815	89	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	53	100	100	89
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	65	100	100	97
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	36	100	92	67
ODC1	{Colonic adenoma recurrence, reduced risk of}, 114500	165640	52	100	100	98
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	42	100	97	81
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	44	100	92	71
ORC1	Meier-Gorlin syndrome 1, 224690	601902	59	100	98	87
OTC	Ornithine transcarbamylase deficiency, 311250	300461	46	100	98	78
P2RX6	No OMIM phenotype	608077	75	97	89	89
PACS1	Schuurs-Hoeijmakers syndrome, 615009	607492	58	100	97	88
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	62	100	98	88
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	45	100	97	82
PAK3	Mental retardation 30/47, 300558	300142	41	100	94	69
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	57	100	100	92
PANX1	No OMIM phenotype	608420	52	100	94	77
PAX1	?Otofaciocervical syndrome 2, 615560	167411	94	100	94	90

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	50	100	98	84
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	167415	76	100	97	93
PC	Pyruvate carboxylase deficiency, 266150	608786	90	100	100	100
PCCA	Propionicacidemia, 606054	232000	50	100	99	85
PCCB	Propionicacidemia, 606054	232050	53	100	96	86
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	92	100	99	92
PCGF2	No OMIM phenotype	600346	65	100	99	95
PCLO	?Pontocerebellar hypoplasia, type 3, 608027	604918	67	100	99	96
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	84	100	99	95
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799	600129	50	100	96	81
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	42	99	93	79
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	72	100	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	50	100	93	84
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	48	100	95	78
PEPD	Prolidase deficiency, 170100	613230	58	100	98	91
PET100	Mitochondrial complex IV deficiency, 220110	614770	75	100	100	81
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	49	100	98	84
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	64	100	96	89
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	105	100	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	54	100	100	86
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	55	100	100	93
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	74	100	95	91
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	46	100	100	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	52	100	100	99
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	76	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	44	100	99	84
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	76	100	100	98
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	62	100	99	91
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	43	100	100	87
PGAP1	Mental retardation 42, 615802	611655	51	100	95	78
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	94	100	100	99
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	62	100	100	98
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	48	100	100	91
PHC1	?Microcephaly 11, primary, 615414	602978	127	100	100	99
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	48	100	95	73
PHF8	Mental retardation syndrome, Siderius type, 300263	300560	49	100	96	74
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	103	100	100	100
PHIP	No OMIM phenotype	612870	53	99	95	80
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	600286	76	100	99	91
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	65	100	100	99
PIGC	No OMIM phenotype	601730	118	100	100	100
PIGG	Mental retardation 53, 616917	616918	75	100	100	99
PIGL	CHIME syndrome, 280000	605947	62	100	100	81
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	45	100	93	72
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	83	100	100	99
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	88	100	100	100
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	71	100	100	94
PIGW	?Hyperphosphatasia with mental retardation syndrome 5, 616025	610275	74	100	100	100
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	40	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
PIK3CA	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000	171834	65	100	99	92
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	59	96	93	89
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	80	100	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	44	100	96	81
PLK4	Microcephaly and chorioretinopathy, 2, 616171	605031	49	100	96	81
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	74	100	99	87
PLXNA3	No OMIM phenotype	300022	67	98	97	95
PLXND1	No OMIM phenotype	604282	77	100	99	96
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	53	100	100	91
PMPCA	Spinocerebellar ataxia 2, 213200	613036	92	100	100	100
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	72	100	100	97
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	54	100	97	89
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	55	100	100	98
POGZ	White-Sutton syndrome, 616364	614787	65	100	98	90
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	71	100	100	99
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	60	100	99	89
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	53	100	97	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	63	100	100	95
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830	614828	119	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	66	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	69	100	100	98
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	52	100	99	89
PORCN	Focal dermal hypoplasia, 305600	300651	74	100	99	93
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	66	100	100	99
POU3F3	No OMIM phenotype	602480	42	72	58	44
PPFIA4	No OMIM phenotype	603145	57	100	98	91
PPM1D	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450	605100	67	100	100	96
PPOX	Porphyria variegata, 176200	600923	73	100	100	98
PPP1CB	No OMIM phenotype	600590	43	100	99	81
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2, 616817	613257	70	100	100	96
PPP2R1A	Mental retardation 36, 616362	605983	86	100	100	100
PPP2R5C	No OMIM phenotype	601645	42	99	86	66
PPP2R5D	Mental retardation 35, 616355	601646	67	100	99	93
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	51	100	100	87
PQBP1	Renpenning syndrome, 309500	300463	92	100	100	100
PRIM1	No OMIM phenotype	176635	37	100	88	61
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	64	100	100	94
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157	610087	69	100	99	94
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	60	100	92	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	43	100	96	79
PRSS12	Mental retardation 1, 249500	606709	59	100	100	93
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	77	100	97	92
PSMD12	No OMIM phenotype	604450	35	98	82	58
PTCH1	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828	601309	70	100	97	92
PTCHD1	{Autism, susceptibility to 4}, 300830	300828	59	100	100	98
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	44	100	99	84
PTEN	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 {Glioma susceptibility 2}, 613028 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 {Meningioma}, 607174 PTEN hamartoma tumor syndrome {Prostate cancer, somatic}, 176807 Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950	601728	85	92	81	77
PTF1A	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069	607194	78	100	100	97
PTGR1	No OMIM phenotype	601274	36	97	83	56
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	57	100	96	80
PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263	608625	63	100	100	99
PTRHD1	No OMIM phenotype	617342	80	100	100	99
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	54	100	100	79

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PUF60	Verheij syndrome, 615583	604819	87	100	100	98
PURA	Mental retardation 31, 616158	600473	126	100	100	100
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	74	100	99	92
PUS3	?Mental retardation 55, 617051	616283	53	100	98	93
PUS7	No OMIM phenotype	616261	40	100	94	73
PYCR1	Cutis laxa, type IIB, 612940 Cutis laxa, type IIIB, 614438	179035	66	100	100	95
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	74	100	100	100
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	75	100	100	96
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	52	100	97	84
RAB18	Warburg micro syndrome 3, 614222	602207	71	100	100	88
RAB27A	Griscelli syndrome, type 2, 607624	603868	37	100	94	71
RAB39B	Mental retardation 72, 300271 ?Waisman syndrome, 311510	300774	50	100	100	96
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	49	100	99	89
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	49	100	96	79
RAB40AL	No OMIM phenotype	300405	173	100	100	100
RAC1	No OMIM phenotype	602048	67	100	100	88
RAD21	Cornelia de Lange syndrome 4, 614701	606462	53	100	99	83
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	71	100	100	95
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	54	100	95	87
RAI1	Smith-Magenis syndrome, 182290	607642	99	100	100	100
RARB	Microphthalmia, syndromic 12, 615524	180220	54	100	100	92
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	47	100	97	76
RBBP8	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744	604124	44	100	98	80
RBFOX1	No OMIM phenotype	605104	56	100	100	90
RBM10	TARP syndrome, 311900	300080	61	100	95	91
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	47	100	99	85
RBPJ	Adams-Oliver syndrome 3, 614814	147183	59	100	95	78
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175	607867	63	100	100	90
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	52	100	98	85
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975	605226	57	98	93	85

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
REV3L	No OMIM phenotype	602776	52	100	99	89
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	43	100	96	79
RHEB	No OMIM phenotype	601293	21	88	54	23
RIT1	Noonan syndrome 8, 615355	609591	53	100	100	94
RLIM	Mental retardation 61, 300978	300379	64	100	100	91
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	50	100	96	70
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	73	100	100	99
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	48	100	97	81
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	139	100	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	68	100	100	84
RNF113A	?Trichothiodystrophy 5, nonphotosensitive, 300953	300951	83	100	100	100
RNF125	Tenorio syndrome, 616260	610432	57	100	100	98
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	59	100	100	97
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	76	100	97	94
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	47	97	94	77
RPL10	{Autism, susceptibility to 5}, 300847	312173	82	100	100	100
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation 19, 300844	300075	38	100	85	57
RSPRY1	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723	616585	53	100	92	75
RTEL1	Dyskeratosis congenita 4, 615190 Dyskeratosis congenita 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	80	100	100	98
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833	610436	49	100	95	79
RUBCN	?Spinocerebellar ataxia 15, 615705	613516	56	100	96	88
RUSC2	No OMIM phenotype	611053	95	100	100	99
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480	602218	99	100	100	100
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	49	100	93	69
SATB2	Glass syndrome, 612313	608148	69	100	98	87
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	49	100	100	92
SC5D	Lathosterolosis, 607330	602286	68	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
SCAF4	No OMIM phenotype	616023	47	100	94	78
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6, 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634	182389	67	100	100	94
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	105	100	93	93
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	73	100	99	95
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	72	100	99	91
SCO1	Mitochondrial complex IV deficiency, 220110	603644	61	100	100	92
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	81	100	100	100
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	93	100	93	83
SEMA3E	?CHARGE syndrome, 214800	608166	44	100	98	78
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	49	100	96	77
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	44	100	92	66
SETBP1	Mental retardation 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	70	98	96	94
SETD1A	No OMIM phenotype	611052	71	99	96	93
SETD2	Luscan-Lumish syndrome, 616831	612778	55	100	99	91
SETD5	Mental retardation 23, 615761	615743	57	100	99	90
SF1	No OMIM phenotype	601516	50	93	87	81
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	90	100	96	94
SH3KBP1	No OMIM phenotype	300374	38	98	86	55
SHANK2	{Autism susceptibility 17}, 613436	603290	87	100	98	95
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	62	93	78	72

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	68	100	98	95
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	48	100	98	85
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434	300579	61	100	98	96
SIK1	Epileptic encephalopathy, early infantile, 30, 616341	605705	84	100	100	100
SIL1	Marinesco-Sjogren syndrome, 248800	608005	54	100	100	93
SIN3A	Witteveen-Kolk syndrome, 613406	607776	50	100	99	88
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	99	100	100	100
SKI	Shprintzen-Goldberg syndrome, 182212	164780	76	100	100	99
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	46	100	97	83
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905	608305	78	100	98	92
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	60	100	100	86
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	60	100	99	83
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	54	100	100	94
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	64	100	99	90
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105	600300	44	100	98	83
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	600229	75	100	100	99
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	61	100	100	91
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	85	100	100	94
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	70	100	100	97
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	81	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	94	100	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	55	100	97	78
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	49	100	99	82
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	55	100	100	98
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	55	100	96	76

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	89	100	100	99
SLC39A12	No OMIM phenotype	608734	42	100	92	72
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721	608732	50	100	97	81
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	43	100	99	83
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	73	100	100	96
SLC6A17	Mental retardation 48, 616269	610299	73	100	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	65	100	100	99
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	79	100	97	92
SLC7A7	Lysinuric protein intolerance, 222700	603593	54	100	97	88
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	54	100	94	82
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	59	100	100	96
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	61	98	96	85
SMARCA4	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325	603254	76	100	100	98
SMARCB1	Coffin-Siris syndrome 3, 614608 {Rhabdoid predisposition syndrome 1}, 609322 Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091	601607	71	100	100	95
SMARCC2	No OMIM phenotype	601734	56	100	99	89
SMARCE1	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174	603111	49	100	100	92
SMC1A	Cornelia de Lange syndrome 2, 300590	300040	59	100	98	95
SMC3	Cornelia de Lange syndrome 3, 610759	606062	56	100	97	83
SMOC1	Microphthalmia with limb anomalies, 206920	608488	68	100	98	91
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	87	100	99	97
SMS	Mental retardation, Snyder-Robinson type, 309583	300105	41	95	80	70
SNAP25	?Myasthenic syndrome, congenital, 18, 616330	600322	59	100	98	88
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	91	100	100	96
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	64	100	100	97
SNRNP70	No OMIM phenotype	180740	79	100	99	96
SNRPN	Prader-Willi syndrome, 176270	182279	64	100	100	100
SNX14	Spinocerebellar ataxia 20, 616354	616105	50	100	97	81
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	64	98	95	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SON	ZTTK syndrome, 617140	182465	70	100	95	90
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	57	100	98	84
SOS2	Noonan syndrome 9, 616559	601247	56	100	98	87
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	68	100	100	100
SOX11	Mental retardation, 27, 615866	600898	102	100	100	100
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	100	100	100	100
SOX3	Mental retardation, with isolated growth hormone deficiency, 300123 Panhypopituitarism, 312000	313430	56	100	96	89
SOX5	Lamb-Shaffer syndrome, 616803	604975	58	100	99	88
SPAST	Spastic paraplegia 4, 182601	604277	46	100	98	80
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577	613940	48	100	98	85
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	56	100	98	92
SPOCK1	No OMIM phenotype	602264	73	100	98	89
SPRED1	Legius syndrome, 611431	609291	44	100	95	84
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	64	100	99	94
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia 14, 615386	604985	73	100	100	100
SRCAP	Floating-Harbor syndrome, 136140	611421	84	100	100	99
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	63	100	99	92
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	47	100	97	85
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	53	100	100	86
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation 12, 611090	606494	52	100	98	87
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	40	94	84	61
STAG1	No OMIM phenotype	604358	35	100	90	62
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	51	100	100	90
STIL	Microcephaly 7, primary, 612703	181590	53	100	99	90
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	69	100	100	99
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	67	100	100	92
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	43	100	99	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	64	100	99	90
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172	601485	81	100	99	91
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	53	100	99	86
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	46	100	94	80
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	61	100	99	93
SUCO	No OMIM phenotype	No id	50	100	99	90
SUMF1	Multiple sulfatase deficiency, 272200	607939	60	100	98	81
SUOX	Sulfite oxidase deficiency, 272300	606887	106	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	59	93	87	87
SUZ12	No OMIM phenotype	606245	54	100	97	87
SYN1	Epilepsy, with variable learning disabilities and behavior disorders, 300491 dominant	313440	58	100	100	97
SYNCRIP	No OMIM phenotype	616686	45	97	87	69
SYNE1	Emery-Dreifuss muscular dystrophy 4, 612998 Spinocerebellar ataxia 8, 610743	608441	55	100	99	89
SYNGAP1	Mental retardation 5, 612621	603384	93	98	98	97
SYP	Mental retardation 96, 300802	313475	50	100	98	87
SYT1	No OMIM phenotype	185605	64	100	100	94
SYT14	Spinocerebellar ataxia 11, 614229	610949	47	100	93	80
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	77	100	100	98
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	48	100	95	83
TAF2	Mental retardation 40, 615599	604912	47	100	95	76
TAT	Tyrosinemia, type II, 276600	613018	48	100	100	91
TBC1D20	Warburg micro syndrome 4, 615663	611663	53	98	93	93
TBC1D24	DOOR syndrome, 220500 Deafness 86, 614617 Deafness 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	87	100	100	98
TBC1D7	Macrocephaly/megalencephaly syndrome, 248000	612655	39	100	93	69
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	604649	60	100	97	87
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	44	98	90	70
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	616899	39	100	93	67

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TBL1XR1	Mental retardation 41, 616944 Pierpont syndrome, 602342	608628	56	100	94	87
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	46	100	99	81
TBR1	No OMIM phenotype	604616	69	100	100	97
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430	602054	44	91	78	67
TCF12	Craniosynostosis 3, 615314	600480	50	100	99	87
TCF20	No OMIM phenotype	603107	83	100	100	100
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954	602272	50	100	97	87
TCF7L2	{Diabetes mellitus, type 2, susceptibility to}, 125853	602228	54	100	98	85
TCN2	Transcobalamin II deficiency, 275350	613441	76	100	100	99
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	50	100	99	90
TDP2	Spinocerebellar ataxia 23, 616949	605764	80	100	100	92
TECPR2	Spastic paraplegia 49, 615031	615000	66	100	99	94
TECR	Mental retardation 14, 614020	610057	81	100	100	94
TELO2	You-Hoover-Fong syndrome, 616954	611140	72	100	100	96
TFAP2A	Branchiooculofacial syndrome, 113620	107580	80	100	100	100
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	96	95	93	92
TGFBR2	No OMIM phenotype	190182	124	100	100	100
TGIF1	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	602630	94	100	100	100
TH	Holoprosencephaly 4, 142946	191290	66	100	100	97
THOC2	Segawa syndrome, recessive, 605407	300395	41	100	95	76
THOC6	Mental retardation 12/35, 300957	615403	133	100	100	100
THRB	Beaulieu-Boycott-Innes syndrome, 613680	190160	56	100	99	93
TIMM8A	Thyroid hormone resistance, 188570 Thyroid hormone resistance, 274300 Thyroid hormone resistance, selective pituitary, 145650	300356	102	100	100	100
TINF2	Mohr-Tranebjaerg syndrome, 304700	604319	94	100	100	100
TLK2	Dyskeratosis congenita 3, 613990 Revesz syndrome, 268130	608439	41	95	77	54

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TM4SF20	No OMIM phenotype	615404	42	100	98	86
TMCO1	{Specific language impairment 5}, 615432	614123	76	100	99	86
TMEM165	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614726	73	100	100	97
TMEM231	Congenital disorder of glycosylation, type IIk, 614727	614949	71	100	100	90
TMEM237	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614423	38	100	90	69
TMEM240	Joubert syndrome 14, 614424	616101	65	100	100	100
TMEM5	Spinocerebellar ataxia 21, 607454	605862	58	100	98	78
TMEM67	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	609884	58	100	98	82
TMEM70	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	612418	78	100	100	94
TMLHE	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	300777	32	100	97	59
TMPRSS7	{Autism, susceptibility to 6}, 300872	617218	36	100	97	72
TMTC3	Lissencephaly 8, 617255	617218	58	100	98	87
TMX2	No OMIM phenotype	616715	48	100	93	79
TNIK	Mental retardation 54, 617028	610005	39	100	96	76
TOE1	Pontocerebellar hypoplasia, type 7, 614969	613931	74	100	100	96
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	106	100	100	98
TPO	Thyroid dyshormonogenesis 2A, 274500	606765	70	100	100	98
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	70	100	100	99
TRAIP	Seckel syndrome 9, 616777	605958	61	100	100	97
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	44	100	95	77
TRAPPC9	Mental retardation 13, 613192	611966	68	100	99	92
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	116	100	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	86	100	100	100
TRIO	Mental retardation 44, 617061	601893	62	99	98	91
TRIP12	No OMIM phenotype	604506	57	100	97	88
TRMT1	No OMIM phenotype	611669	65	100	100	96
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	616013	67	100	99	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	105	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	104	100	100	100
TSEN15	Pontocerebellar hypoplasia, type 2F, 617026	608756	62	100	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	100	96	95
TSHB	Hypothryoidism, congenital, nongoitrous 4, 275100	188540	70	100	100	100
TSPAN7	Mental retardation 58, 300210	300096	45	100	95	77
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	39	100	89	62
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	43	100	97	80
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	46	100	99	85
TTI2	Mental retardation 39, 615541	614426	46	100	98	81
TUBA1A	Lissencephaly 3, 611603	602529	89	100	100	100
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180	605742	79	100	100	100
TUBB	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610	191130	135	100	100	99
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	170	100	97	95
TUBB2B	Polymicrogyria, symmetric or asymmetric, 610031	612850	170	100	100	100
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	168	100	95	95
TUBB4A	Dystonia 4, torsion, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	148	100	100	97
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	99	100	100	100
TUBGCP4	Microcephaly and chorioretinopathy, 3, 616335	609610	51	100	96	80
TUBGCP6	Microcephaly and chorioretinopathy, 1, 251270	610053	109	100	100	99
TUSC3	Mental retardation 7, 611093	601385	55	100	99	88
TWIST1	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400	601622	72	100	90	77
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	109	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
UBA5	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia 24, 617133	610552	42	100	96	75
UBE2A	Mental retardation syndromic, Nascimento-type, 300860	312180	56	100	88	75
UBE3A	Angelman syndrome, 105830	601623	53	100	100	93
UBE3B	Kaufman oculocerebrofacial syndrome, 244450	608047	72	100	100	93
UBR1	Johanson-Blizzard syndrome, 243800	605981	43	100	97	77
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801	612636	52	100	99	89
UPB1	Beta-ureidopropionase deficiency, 613161	606673	65	100	100	96
UPF3B	Mental retardation, syndromic 14, 300676	300298	61	100	97	89
UQCC2	?Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	80	100	100	89
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	86	100	100	100
UROC1	?Urocanase deficiency, 276880	613012	59	100	100	97
USP18	Pseudo-TORCH syndrome 2, 617397	607057	85	100	95	95
USP27X	Mental retardation 105, 300984	300975	71	100	100	97
USP7	No OMIM phenotype	602519	33	97	82	55
USP9X	Mental retardation 99, 300919 Mental retardation 99, syndromic, female-restricted, 300968	300072	63	100	98	94
UTRN	No OMIM phenotype	128240	55	100	98	88
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	48	100	99	87
VPS11	Leukodystrophy, hypomyelinating, 12, 616683	608549	67	100	99	94
VPS13B	Cohen syndrome, 216550	607817	54	100	98	87
VPS13C	Parkinson disease 23, early onset, 616840	608879	38	100	93	70
VPS37A	Spastic paraplegia 53, 614898	609927	39	99	93	68
VPS53	Pontocerebellar hypoplasia, type 2E, 615851	615850	52	100	96	83
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	42	100	98	81
VWA3B	?Spinocerebellar ataxia 22, 616948	614884	46	100	93	77
WAC	Desanto-Shinawi syndrome, 616708	615049	42	100	92	72
WASHC4	?Mental retardation 43, 615817	615748	54	100	96	79
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	45	100	97	78
WDR13	No OMIM phenotype	300512	64	100	100	96
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	50	100	98	85
WDR4	No OMIM phenotype	605924	63	100	100	94
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	87	100	100	100
WDR54	No OMIM phenotype	No id	77	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
WDR62	Microcephaly 2, primary, with or without cortical malformations, 604317	613583	79	100	100	98
WDR73	Galloway-Mowat syndrome, 251300	616144	83	100	97	91
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185	614218	96	100	100	100
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome, 222300 Wolfram-like syndrome, 614296	606201	106	100	100	100
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia 12, 614322	605131	57	100	100	93
XPA	Xeroderma pigmentosum, group A, 278700	611153	45	100	92	70
XPC	Xeroderma pigmentosum, group C, 278720	613208	66	100	98	90
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	41	100	98	82
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	74	98	92	88
YAP1	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	49	100	95	79
YME1L1	?Optic atrophy 11, 617302	607472	36	100	91	64
YWHAE	No OMIM phenotype	605066	50	100	99	89
YY1	No OMIM phenotype	600013	74	100	100	98
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	92	100	100	99
ZBTB18	Mental retardation 22, 612337	608433	89	100	99	98
ZBTB20	Primrose syndrome, 259050	606025	123	100	100	100
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069	614064	57	100	100	99
ZC3H14	Mental retardation 56, 617125	613279	65	100	98	88
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	62	100	100	88
ZDHHC15	?Mental retardation 91, 300577	300576	39	100	96	70
ZDHHC9	Mental retardation syndromic, Raymond type, 300799	300646	39	100	95	69
ZEB2	Mowat-Wilson syndrome, 235730	605802	61	100	100	98
ZFYVE26	Spastic paraplegia 15, 270700	612012	64	100	99	91
ZIC1	Craniosynostosis 6, 616602	600470	110	100	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	82	95	90	87
ZIC4	No OMIM phenotype	608948	92	100	100	97
ZMYND11	Mental retardation 30, 616083	608668	53	100	99	90
ZNF101	No OMIM phenotype	603983	46	100	100	88
ZNF292	No OMIM phenotype	616213	51	100	97	93

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ZNF407	No OMIM phenotype	615894	68	100	100	98
ZNF41	No OMIM phenotype	314995	48	100	98	90
ZNF592	No OMIM phenotype	613624	87	100	100	100
ZNF674	No OMIM phenotype	300573	59	100	100	100
ZNF711	Mental retardation 97, 300803	314990	50	100	93	83
ZNF81	No OMIM phenotype	314998	46	100	100	87
ZSWIM6	Acromelic frontonasal dysostosis, 603671	615951	46	95	90	80

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "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 96 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 describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x