Whole Exome Sequencing
Gene package Intellectual disability, version 5, 1-7-2017

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
A2ML1	No OMIM phenotype	610627	44	99	90
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	86	100	100
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	77	100	99
ABAT	GABA-transaminase deficiency, 613163	137150	77	100	99
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	81	100	98
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	46	96	86
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	88	100	98
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	100	100	100
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	88	100	100
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	91	98	92
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	93	100	100
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	82	100	100
ACSL4	Mental retardation 63, 300387	300157	44	99	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	77	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	104	100	100
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	86	100	100
ACY1	Aminoacylase 1 deficiency, 609924	104620	84	100	100
ADA2	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	34	93	73
ADAM22	No OMIM phenotype	603709	78	100	100
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	96	100	100
ADAT3	Mental retardation 36, 615286	615302	53	99	95
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	78	100	100
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	60	99	92
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	79	100	99
ADSL	Adenylosuccinase deficiency, 103050	608222	86	100	100
AFF2	Mental retardation, FRAXE type, 309548	300806	45	100	93
AFF4	CHOPS syndrome, 616368	604417	44	96	85
AFG3L2	Spastic ataxia 5, 614487 Spinocerebellar ataxia 28, 610246	604581	68	90	84
AGA	Aspartylglucosaminuria, 208400	613228	79	100	100
AGAP2	No OMIM phenotype	605476	55	98	88
AGO2	No OMIM phenotype	606229	47	98	91
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	72	100	100
AGTR2	No OMIM phenotype	300034	74	100	100
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	96	100	100
AHDC1	Xia-Gibbs syndrome, 615829	615790	63	99	93
AHI1	Joubert syndrome 3, 608629	608894	77	100	98
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness 5, 300614	300169	55	100	94
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	66	100	98
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	98	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	87	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	33	89	66
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	82	100	99
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	91	100	100
ALDH4A1	Hyperprolinemia, type II, 239510	606811	66	100	99
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	61	97	87
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	74	100	98
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	72	99	96
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	101	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	30	88	60
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	63	100	100
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	63	100	98
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	65	100	98
ALG8	Congenital disorder of glycosylation, type Ih, 608104	608103	56	97	90
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	68	100	97
ALMS1	Alstrom syndrome, 203800	606844	105	100	100
ALX1	?Frontonasal dysplasia 3, 613456	601527	96	100	100
ALX4	{Craniosynostosis 5, susceptibility to}, 615529 Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597	605420	82	100	100
AMMECR1	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990	300195	27	97	78
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686	102771	60	100	94
AMT	Glycine encephalopathy, 605899	238310	91	100	100
ANK3	?Mental retardation, 37, 615493	600465	72	100	99
ANKEF1	No OMIM phenotype	No id	53	99	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ANKH	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	605145	79	100	100
ANKLE2	?Microcephaly 16, primary, 616681	616062	64	98	90
ANKRD11	KBG syndrome, 148050	611192	77	100	97
ANO10	Spinocerebellar ataxia 10, 613728	613726	73	100	100
ANTXR1	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089	606410	78	98	91
AP1S1	MEDNIK syndrome, 609313	603531	77	100	99
AP1S2	Mental retardation syndromic 5, 304340	300629	32	97	79
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	73	100	95
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276	602166	86	97	93
AP4B1	Spastic paraplegia 47, 614066	607245	83	100	100
AP4E1	Spastic paraplegia 51, 613744 Stuttering, familial persistent, 1, 184450	607244	61	100	98
AP4M1	Spastic paraplegia 50, 612936	602296	75	100	100
AP4S1	Spastic paraplegia 52, 614067	607243	40	97	85
APC2	?Sotos syndrome 3, 617169	612034	44	93	77
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	86	100	98
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	86	100	100
ARG1	Argininemia, 207800	608313	71	100	100
ARHGAP4	No OMIM phenotype	300023	30	96	69
ARHGEF6	Mental retardation 46, 300436	300267	50	99	88
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	47	100	87
ARID1A	Coffin-Siris syndrome 2, 614607	603024	83	97	94
ARID1B	Coffin-Siris syndrome 1, 135900	614556	83	98	93
ARID2	No OMIM phenotype	609539	62	96	87
ARL13B	Joubert syndrome 8, 612291	608922	61	100	100
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	48	95	81
ARNT2	?Webb-Dattani syndrome, 615926	606036	51	100	92
ARSA	Metachromatic leukodystrophy, 250100	607574	79	100	100
ARSE	Chondrodysplasia punctata recessive, 302950	300180	44	94	77

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	24	84	61
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	77	100	99
ASCL1	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880	100790	117	100	100
ASL	Argininosuccinic aciduria, 207900	608310	72	100	100
ASNS	Asparagine synthetase deficiency, 615574	108370	64	93	84
ASPA	Canavan disease, 271900	608034	85	100	97
ASPM	Microcephaly 5, primary, 608716	605481	62	100	96
ASS1	Citrullinemia, 215700	603470	111	100	100
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	77	100	98
ASXL2	Shashi-Pena syndrome, 617190	612991	61	98	97
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	53	99	94
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	99	100	100
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	71	100	99
ATN1	Dentatorubro-pallidoluysian atrophy, 125370	607462	79	100	99
ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	109	100	100
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	96	100	99
ATP2A2	Acrokeratosis verruciformis, 101900 Darier disease, 124200	108740	95	100	100
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	39	100	90
ATP6V0A2	Cutis laxa, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	94	100	100
ATP6V1B2	Deafness, congenital, with onychodystrophy, 124480 Zimmermann-Laband syndrome 2, 616455	606939	47	98	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	53	99	94
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	82	100	98
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	76	100	100
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600	601215	81	100	98
ATRIP	No OMIM phenotype	606605	59	99	91
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	38	98	83
AUH	3-methylglutaconic aciduria, type I, 250950	600529	60	100	90
AUTS2	Mental retardation 26, 615834	607270	81	99	93
AVPR2	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	300538	47	100	97
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	83	100	97
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	38	75	71
B3GLCT	Peters-plus syndrome, 261540	610308	65	98	90
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	70	100	100
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070	604327	87	97	95
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	76	100	99
BBS1	Bardet-Biedl syndrome 1, 209900	209901	92	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	76	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	98	100	100
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	76	100	98
BBS4	Bardet-Biedl syndrome 4, 615982	600374	80	100	99
BBS5	Bardet-Biedl syndrome 5, 615983	603650	59	99	92
BBS7	Bardet-Biedl syndrome 7, 615984	607590	68	100	98
BBS9	Bardet-Biedl syndrome 9, 615986	607968	76	98	93
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	22	76	46
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	112	100	99
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	93	100	92
BCL11A	Dias-Logan syndrome, 617101	606557	73	100	100
BCOR	Microphthalmia, syndromic 2, 300166	300485	52	99	94
BCORL1	No OMIM phenotype	300688	58	100	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	114	100	100
BLM	Bloom syndrome, 210900	604610	74	100	99
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	60	98	88
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	69	100	97
BRF1	Cerebellofaciodental syndrome, 616202	604902	46	98	82
BRPF1	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333	602410	70	100	97
BRWD3	Mental retardation 93, 300659	300553	48	98	90
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	82	100	100
BTD	Biotinidase deficiency, 253260	609019	105	100	100
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	72	100	95
c12orf4	No OMIM phenotype	616082	40	95	80
C12orf57	Temtamy syndrome, 218340	615140	85	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, 615035	613541	56	100	100
C1orf167	No OMIM phenotype	No id	41	94	78
C2CD3	?Orofaciodigital syndrome XIV, 615948	615944	47	98	91
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	78	100	96
CA2	Osteopetrosis 3, with renal tubular acidosis, 259730	611492	89	100	100
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	38	94	72
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	66	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	74	100	95
CACNA1C	Brugada syndrome 3, 611875 Timothy syndrome, 601005	114205	96	100	100
CACNA2D1	No OMIM phenotype	114204	55	98	88
CACNG2	?Mental retardation 10, 614256	602911	100	100	100
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	58	98	92
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	100	100	99
CAPN10	{Diabetes mellitus, noninsulin-dependent 1}, 601283	605286	74	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	39	98	87
CBL	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	68	100	100
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	82	100	99
CC2D1A	Mental retardation 3, 608443	610055	74	100	99
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	82	100	99
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	82	100	96
CCDC13	No OMIM phenotype	No id	45	99	89
CCDC14	No OMIM phenotype	617147	68	100	100
CCDC22	Ritscher-Schinzel syndrome 2, 300963	300859	45	99	87
CCDC78	Myopathy, centronuclear, 4, 614807	614666	64	100	100
CCDC88C	Hydrocephalus, nonsyndromic, 236600 ?Spinocerebellar ataxia 40, 616053	611204	77	100	100
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	123833	57	100	98
CDC5L	No OMIM phenotype	602868	39	96	81
CDH15	Mental retardation 3, 612580	114019	84	100	97
CDK16	No OMIM phenotype	311550	48	100	98
CDK5	?Lissencephaly 7 with cerebellar hypoplasia, 616342	123831	45	100	87
CDK5RAP2	Microcephaly 3, primary, 604804	608201	69	100	97
CDK6	?Microcephaly 12, primary, 616080	603368	77	99	95
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	47	98	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	46	80	72
CDON	Holoprosencephaly 11, 614226	608707	90	100	100
CENPJ	Microcephaly 6, primary, 608393 ?Seckel syndrome 4, 613676	609279	80	100	98
CEP104	Joubert syndrome 25, 616781	616690	47	99	88
CEP135	Microcephaly 8, primary, 614673	611423	60	100	97
CEP152	Microcephaly 9, primary, 614852 Seckel syndrome 5, 613823	613529	76	100	98
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	55	98	92
CEP41	Joubert syndrome 15, 614464	610523	64	100	93
CEP63	?Seckel syndrome 6, 614728	614724	69	100	94
CEP89	No OMIM phenotype	615470	54	96	87
CHAMP1	Mental retardation 40, 616579	616327	80	100	100
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	75	100	98
CHD3	No OMIM phenotype	602120	73	97	92
CHD4	Sifrim-Hitz-Weiss syndrome, 617159	603277	46	100	92
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	80	100	98
CHD8	{Autism, susceptibility to, 18}, 615032	610528	83	100	99
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	57	100	99
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	80	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	91	97	94
CIC	No OMIM phenotype	612082	43	63	62
CIT	Microcephaly 17, primary, 617090	605629	40	97	83
CKAP2L	Filippi syndrome, 272440	616174	64	96	91
CLCN4	Mental retardation 49/15, 300114	302910	53	100	100
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	70	98	86
CLIC2	?Mental retardation, syndromic 32, 300886	300138	46	100	100
CLIP1	No OMIM phenotype	179838	47	98	86
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	68	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	73	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	72	100	91
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	100	100	100
CLP1	Pontocerebellar hypoplasia, type 10, 615803	608757	56	100	96
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	58	99	92
CNKSR2	No OMIM phenotype	300724	42	98	90
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	113	100	100
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	85	100	100
COASY	Neurodegeneration with brain iron accumulation 6, 615643	609855	74	100	100
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	91	100	98
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	71	100	99
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	60	100	95
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	72	99	96
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	79	100	100
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	72	100	100
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	56	98	88
COL18A1	Knobloch syndrome, type 1, 267750	120328	71	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	76	99	96
COL4A2	{Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 2, 614483	120090	69	100	97
COL4A3BP	Mental retardation 34, 616351	604677	47	94	82
COLEC11	3MC syndrome 2, 265050	612502	99	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	94
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276	612898	49	100	93
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	95	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	75	100	95
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	105	100	100
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	86	100	100
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	137	100	100
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	78	100	99
CRADD	Mental retardation 34, with variant lissencephaly, 614499	603454	73	100	96
CRB2	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	609720	57	98	89
CRBN	Mental retardation 2, 607417	609262	87	100	94
CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	81	100	97
CRLF1	Cold-induced sweating syndrome 1, 272430	604237	84	91	91
CSNK2A1	Okur-Chung neurodevelopmental syndrome, 617062	115440	43	95	75
CSPP1	Joubert syndrome 21, 615636	611654	38	96	81
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	124	100	97
CTBP1	No OMIM phenotype	602618	43	95	80
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	71	100	100
CTCF	Mental retardation 21, 615502	604167	47	96	89
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	74	95	88
CTNNB1	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	81	100	100
CTNND1	No OMIM phenotype	601045	49	99	92
CTNND2	No OMIM phenotype	604275	76	94	91
CTSA	Galactosialidosis, 256540	613111	81	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	83	100	96
CTTNBP2	No OMIM phenotype	609772	78	100	99
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	83	99	98
CUL4B	Mental retardation, syndromic 15 (Cabezas type), 300354	300304	39	100	92
CWF19L1	Spinocerebellar ataxia 17, 616127	616120	41	97	76
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	86	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	95	99	95
CYP2U1	Spastic paraplegia 56, 615030	610670	89	96	93
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	91	100	100
DAB1	No OMIM phenotype	603448	43	100	92
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	128239	108	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	71	100	97
DBT	Maple syrup urine disease, type II, 248600	248610	70	100	95
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	71	100	98
DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Mirror movements 1, 157600	120470	85	100	97
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	70	99	97
DCPS	Al-Raqad syndrome, 616459	610534	61	100	94
DCX	Lissencephaly, 300067 Subcortical laminal heteropia, 300067	300121	47	98	85
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	95	100	100
DDHD2	Spastic paraplegia 54, 615033	615003	86	100	100
DDX11	Warsaw breakage syndrome, 613398	601150	39	78	66
DDX3X	Mental retardation 102, 300958 dominant	300160	50	99	94
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation 24, 615828	602635	81	87	82
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281	617278	39	99	87
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	53	99	90
DHCR24	Desmosterolosis, 602398	606418	104	100	100
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	114	100	100
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	21	74	44
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	100	100	100
DIAPH1	Deafness 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	80	100	96
DIP2B	Mental retardation, FRA12A type, 136630	611379	83	100	98
DKC1	Dyskeratosis congenita, 305000	300126	57	100	94
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	84	100	97
DLG3	Mental retardation 90, 300850	300189	40	100	84
DLG4	No OMIM phenotype	602887	65	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	50	99	93
DMPK	Myotonic dystrophy 1, 160900	605377	70	100	97
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	62	100	97
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	35	88	60
DNM1	Epileptic encephalopathy, early infantile, 31, 616346	602377	93	98	97
DNMT3A	Tatton-Brown-Rahman syndrome, 615879	602769	72	100	98
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	89	100	99
DOCK6	Adams-Oliver syndrome 2, 614219	614194	73	99	98
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859	615730	71	100	97
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	84	100	99
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	84	100	100
DONSON	No OMIM phenotype	611428	35	91	69
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	80	100	99
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901	603527	69	100	98
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	69	100	100
DPP6	Mental retardation 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956	126141	70	99	94
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	79	98	95
DPYS	Dihydropyrimidinuria, 222748	613326	103	100	98
DST	Epidermolysis bullosa simplex 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653	113810	90	100	98
DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	64	100	98
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	86	100	99
DYRK1A	Mental retardation 7, 614104	600855	99	100	100
EBP	Chondrodysplasia punctata dominant, 302960 MEND syndrome, 300960	300205	43	100	93
EDC3	?Mental retardation 50, 616460	609842	46	99	89
EDNRB	ABCD syndrome, 600501 {Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580	131244	93	100	100
EDRF1	No OMIM phenotype	No id	71	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation 38, 616393	602959	78	100	96
EFCAB1	No OMIM phenotype	No id	36	96	77
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536	603892	84	100	99
EHMT1	Kleefstra syndrome, 610253	607001	97	99	99
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	73	98	91
EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305	608546	35	99	81
EIF4G1	{Parkinson disease 18}, 614251	600495	92	100	99
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731	605367	73	100	100
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	57	100	98
ELP2	Mental retardation 58, 617270	616054	71	100	98
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	45	99	88
EMG1	Bowen-Conradi syndrome, 211180	611531	78	100	100
EML1	Band heterotopia, 600348	602033	46	99	89
EMX2	Schizencephaly, 269160	600035	95	100	100
ENTPD1	Spastic paraplegia 64, 615683	601752	88	100	100
EOMES	No OMIM phenotype	604615	54	100	87
EP300	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684	602700	90	100	100
EPB41L1	?Mental retardation 11, 614257	602879	88	100	99
EPG5	Vici syndrome, 242840	615068	42	96	83
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	73	100	92
ERCC2	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	77	100	100
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	133510	81	100	100
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	133530	73	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	93	100	100
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	60	100	96
ERLIN2	Spastic paraplegia 18, 611225	611605	92	100	100
ERMARD	?Periventricular nodular heterotopia 6, 615544	615532	45	95	78
ESCO2	Roberts syndrome, 268300 SC phocomelia syndrome, 269000	609353	58	98	92
ETFB	Glutaric acidemia IIB, 231680	130410	82	100	100
ETHE1	Ethylmalonic encephalopathy, 602473	608451	67	100	99
EXOSC2	No OMIM phenotype	602238	41	100	86
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	81	100	100
EZH2	Weaver syndrome, 277590	601573	84	100	99
FA2H	Spastic paraplegia 35, 612319	611026	55	98	87
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	67	100	94
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154	616107	27	88	58
FAT2	No OMIM phenotype	604269	58	99	93
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	78	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	86	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	90	100	98
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	605654	64	100	100
FBXO31	?Mental retardation 45, 615979	609102	42	94	83
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation syndromic 16, 305400	300546	43	99	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FGF12	Epileptic encephalopathy, early infantile, 47, 617166	601513	39	99	90
FGF14	Spinocerebellar ataxia 27, 609307	601515	96	100	100
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	99	100	100
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	80	100	98
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	73	100	100
FGL1	No OMIM phenotype	605776	43	99	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	84	97	91
FIBP	Thauvin-Robinet-Faivre syndrome, 617107	608296	52	100	94
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	86	100	99
FIGN	No OMIM phenotype	605295	54	100	98
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	72	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	75	100	100
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	55	100	99
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	82	100	97
FMN2	Mental retardation 47, 616193	606373	72	99	94
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	44	100	89
FOXG1	Rett syndrome, congenital variant, 613454	164874	80	83	77
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	80	100	98
FOXP2	Speech-language disorder-1, 602081	605317	76	100	100
FRAS1	Fraser syndrome, 219000	607830	91	100	100
FREM2	Fraser syndrome, 219000	608945	92	100	100
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	616305	51	99	89
FRMPD4	Mental retardation 104, 300983	300838	48	100	91
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981	604574	38	69	60

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	57	95	90
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	70	100	100
FTSJ1	Mental retardation 9/44, 309549	300499	59	100	100
FUCA1	Fucosidosis, 230000	612280	96	100	100
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	95	100	100
GABRB1	Epileptic encephalopathy, early infantile, 45, 617153	137190	63	100	97
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	94	96	94
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	78	100	100
GALE	Galactose epimerase deficiency, 230350	606953	84	100	100
GALT	Galactosemia, 230400	606999	120	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	59	99	89
GAS6	No OMIM phenotype	600441	47	95	88
GATAD2B	Mental retardation 18, 615074	614998	40	98	82
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	93	100	100
GCDH	Glutaricaciduria, type I, 231670	608801	81	100	99
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	63	100	87
GCSH	Glycine encephalopathy, 605899	238330	14	61	18
GDI1	Mental retardation 41, 300849	300104	64	100	98
GFAP	Alexander disease, 203450	137780	62	100	100
GFM2	No OMIM phenotype	606544	43	94	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	66	100	100
GJB1	Charcot-Marie-Tooth neuropathy dominant, 1, 302800	304040	54	100	100
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, 613206	608803	40	83	58
GK	Glycerol kinase deficiency, 307030	300474	33	99	74

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	77	100	97
GLDC	Glycine encephalopathy, 605899	238300	36	81	62
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	82	99	97
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	88	100	100
GLYCTK	D-glyceric aciduria, 220120	610516	92	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	94	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	57	100	95
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	100	100	100
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	66	100	99
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	600998	35	69	47
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	91	100	97
GNB1	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation 42, 616973	139380	61	100	97
GNB5	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182	604447	45	93	84
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	92	100	100
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	96	100	97
GNS	Mucopolysaccharidosis type IIID, 252940	607664	84	100	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	38	98	92
GPHN	Molybdenum cofactor deficiency C, 615501	603930	80	100	98
GPT2	Mental retardation 49, 616281	138210	55	99	91
GRIA3	Mental retardation 94, 300699	305915	54	100	98
GRID2	Spinocerebellar ataxia 18, 616204	602368	93	100	100
GRIK2	Mental retardation, 6, 611092	138244	88	100	99
GRIN1	Mental retardation 8, 614254	138249	96	100	99
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	80	100	100
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	95	100	100
GRIN3B	No OMIM phenotype	606651	59	85	75
GRIP1	Fraser syndrome, 219000	604597	82	100	100
GRM1	Spinocerebellar ataxia 13, 614831	604473	89	100	100
GSE1	No OMIM phenotype	616886	68	100	97
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	80	100	99
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	112	100	100
GTPBP3	Combined oxidative phosphorylation deficiency 23, 616198	608536	62	100	95
GUCY2F	No OMIM phenotype	300041	42	100	92
GUSB	Mucopolysaccharidosis VII, 253220	611499	95	100	99
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	76	100	98
HAX1	Neutropenia, severe congenital 3, 610738	605998	100	100	100
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	53	100	99
HCFC1	Mental retardation 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	44	100	94
HCN1	Epileptic encephalopathy, early infantile, 24, 615871	602780	70	100	99
HDAC4	No OMIM phenotype	605314	73	100	100
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	300272	53	100	97
HDAC8	Cornelia de Lange syndrome 5, 300882	300269	41	100	94
HECTD1	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	617245	52	96	88
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	617245	48	97	87
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	63	99	83
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011	605109	54	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HERC2	Mental retardation 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	37	73	63
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230	601802	78	100	100
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	85	100	100
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	88	100	95
HIVEP2	Mental retardation 43, 616977	143054	69	100	99
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	88	100	100
HMGCL	HMG-CoA lyase deficiency, 246450	613898	98	100	100
HNMT	{Asthma, susceptibility to}, 600807 Mental retardation 51, 616739	605238	63	100	94
HNRNPH2	Mental retardation, syndromic, Bain type, 300986	300610	40	100	97
HNRNPK	Au-Kline syndrome, 616580	600712	23	76	52
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391	602869	73	100	97
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	108	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	102	100	100
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	36	100	85
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	128	100	100
HSD17B10	HDS10 mitochondrial disease, 300438	300256	63	100	100
HSPA9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854	600548	83	100	99
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, 605280	118190	27	84	58
HUWE1	Mental retardation syndromic, Turner type, 300706	300697	43	99	89
IARS	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093	600709	47	99	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
IBA57	?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, 616451	615316	59	95	90
IDS	Mucopolysaccharidosis II, 309900	300823	62	100	100
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	67	92	82
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	60	78	78
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	77	100	100
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	77	100	99
IFT81	No OMIM phenotype	605489	31	84	57
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	56	100	100
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	84	100	100
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	25	75	49
IL1RAPL1	Mental retardation 21/34, 300143	300206	50	100	100
IMPA1	Mental retardation 59, 617323	602064	28	85	58
INPP5B	No OMIM phenotype	147264	86	100	99
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	71	98	92
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404	607875	40	99	86
INTS1	No OMIM phenotype	611345	50	98	89
INTS8	No OMIM phenotype	611351	40	92	70
IQSEC2	Mental retardation 1/78, 309530	300522	38	94	84
ISG15	Immunodeficiency 38, 616126	147571	71	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	50	97	88
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	600536	78	97	96
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	96	100	100
ITPR2	?Anhidrosis, isolated, with normal sweat glands, 106190	600144	47	96	84
IVD	Isovaleric acidemia, 243500	607036	81	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
JAG1	Alagille syndrome 1, 118450 ?Deafness, congenital heart defects, and posterior embryotoxon Tetralogy of Fallot, 187500	601920	89	99	96
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	102	100	100
JMJD1C	No OMIM phenotype	604503	69	100	98
KALRN	{Coronary heart disease, susceptibility to, 5}, 608901	604605	89	100	100
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900	607704	81	100	100
KANSL1	Koolen-De Vries syndrome, 610443	612452	65	97	86
KAT6A	Mental retardation 32, 616268	601408	82	100	98
KAT6B	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736	605880	94	100	99
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	67	100	98
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366	176262	58	100	100
KCNA4	No OMIM phenotype	176266	81	100	100
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056	600397	60	99	96
KCNC3	Spinocerebellar ataxia 13, 605259	176264	56	80	66
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500	603305	65	99	95
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	122	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	104	100	100
KCNJ6	Keppen-Lubinsky syndrome, 614098	600877	80	100	100
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292	605874	88	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	73	100	97
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data		
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200	602235	70	100	100
KCNQ3	Seizures, benign neonatal, type 2, 121201	602232	85	100	96
KCNQ5	No OMIM phenotype	607357	76	98	95
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959	608167	86	100	99
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	90	100	100
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features, 616728	609132	51	99	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KDM4B	No OMIM phenotype	609765	60	99	92
KDM5C	Mental retardation, syndromic, Claes-Jensen type, 300534	314690	52	100	96
KDM6A	Kabuki syndrome 2, 300867	300128	36	95	84
KDSR	Lymphoma/leukemia, B-cell, variant	136440	53	100	90
KIAA1109	No OMIM phenotype	611565	49	96	86
KIAA1586	No OMIM phenotype	No id	31	92	66
KIDINS220	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296	615759	54	99	91
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	53	99	95
KIF1A	Mental retardation 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, 610357	601255	75	100	98
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	90	100	95
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	36	87	63
KIF4A	?Mental retardation 100, 300923	300521	23	87	57
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282	604593	42	96	79
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	64	98	93
KIRREL3	Mental retardation 4, 612581	607761	77	100	99
KLHL15	Mental retardation 103, 300982	300980	40	99	91
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	55	99	95
KMT2B	Dystonia 28, childhood-onset, 617284	606834	62	93	88
KMT2C	No OMIM phenotype	606833	56	89	84
KMT2D	Kabuki syndrome 1, 147920	602113	80	100	99
KNL1	Microcephaly 4, primary, 604321	609173	55	99	91
KPTN	Mental retardation 41, 615637	615620	54	100	95
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	64	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KRBOX4	No OMIM phenotype	300585	59	100	100
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	52	100	100
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	64	100	92
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	88	100	99
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855	156225	75	100	99
LAMC1	No OMIM phenotype	150290	55	99	94
LAMC3	Cortical malformations, occipital, 614115	604349	88	100	98
LAMP2	Danon disease, 300257	309060	65	91	90
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	91	100	100
LARP7	Alazami syndrome, 615071	612026	23	63	41
LAS1L	Wilson-Turner syndrome, 309585	300964	41	99	88
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462	607031	85	100	97
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	76	100	100
LINS1	Mental retardation 27, 614340	610350	79	100	100
LMAN2L	?Mental retardation, 52, 616887	609552	48	97	91
LONP1	CODAS syndrome, 600373	605490	64	99	97
LRP2	Donnai-Barrow syndrome, 222448	600073	89	100	100
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	76	100	98
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	87	100	92
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	96	100	100
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	53	81	74
MAGEC3	No OMIM phenotype	300469	25	91	66
MAGEL2	Schaaf-Yang syndrome, 615547	605283	70	100	97
MAGT1	Immunodeficiency, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	44	100	89
MAN1B1	Mental retardation 15, 614202	604346	91	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	86	100	98
MANBA	Mannosidosis, beta, 248510	609489	84	100	100
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	39	100	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	87	100	100
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	91	100	100
MAPRE2	Symmetric circumferential skin creases, congenital, 2, 616734	605789	62	98	90
MASP1	3MC syndrome 1, 257920	600521	103	100	99
MAT1A	Hypermethioninemia, persistent, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, 250850	610550	89	100	100
MBD5	Mental retardation 1, 156200	611472	82	100	100
MBOAT7	Mental retardation 57, 617188	606048	56	100	96
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, 308800 ?Olmsted syndrome, 300918	300294	54	100	99
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	77	100	100
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	62	100	95
MCM3AP	No OMIM phenotype	603294	53	99	92
MCOLN1	Mucolipidosis IV, 252650	605248	95	100	99
MCOLN3	No OMIM phenotype	607400	37	94	78
MCPH1	Microcephaly 1, primary, 251200	607117	67	100	99
MDH2	Epileptic encephalopathy, early infantile, 51, 617339	154100	42	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	52	99	81
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	45	98	87
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, 300895 Opitz-Kaveggia syndrome, 305450	300188	47	100	91
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808	608771	77	100	99
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	77	100	100
MED23	Mental retardation 18, 614249	605042	72	100	97
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589	610197	77	100	98
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4) Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	66	100	98
MEIS2	Cleft palate, cardiac defects, and mental retardation, 600987	601740	47	100	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
METTL23	Mental retardation 44, 615942	615262	42	100	100
MFSD2A	Microcephaly 15, primary, 616486	614397	48	99	88
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	75	100	97
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	87	100	100
MICU1	Myopathy with extrapyramidal signs, 615673	605084	37	93	80
MID1	Opitz GBBB syndrome, type I, 300000	300552	57	100	99
MID2	?Mental retardation 101, 300928	300204	33	96	76
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	106	100	100
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	73	100	98
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	71	100	96
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	67	95	92
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	105	100	100
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	135	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	60	100	97
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	67	93	77
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	73	100	100
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	78	100	100
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	66	100	99
MPDZ	Hydrocephalus, nonsyndromic 2, 615219	603785	76	100	97
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	77	100	100
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582	607118	43	79	63
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	75	100	99
MSL2	No OMIM phenotype	614802	82	100	100
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947	611766	67	100	95
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	94	100	100
MTMR9	No OMIM phenotype	606260	24	97	66
MTOR	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638	601231	47	98	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	80	100	99
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	81	100	100
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	81	100	100
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	94	100	100
MYCN	Feingold syndrome 1, 164280	164840	68	100	91
MYH9	Deafness 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249	160775	91	100	100
MYO5A	Griscelli syndrome, type 1, 214450	160777	74	100	98
MYT1L	Mental retardation 39, 616521	613084	89	100	99
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	51	100	100
NAA15	No OMIM phenotype	608000	32	91	68
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393	610672	73	100	97
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	86	100	100
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	81	100	93
NALCN	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	611549	43	98	87
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	36	99	87
NARS2	Combined oxidative phosphorylation deficiency 24, 616239	612803	67	97	97
NAT10	No OMIM phenotype	609221	52	99	95
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	66	100	97
NCOR2	No OMIM phenotype	600848	42	95	80
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	94	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	68	100	100
NDST1	Mental retardation 46, 616116	600853	109	100	100
NDUFA1	Mitochondrial complex I deficiency, 252010 dominant	300078	85	100	90
NDUFA11	Mitochondrial complex I deficiency, 252010 dominant	612638	57	100	96
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000	614530	98	100	100
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	71	100	100
NDUFAF3	Mitochondrial complex I deficiency, 252010 dominant	612911	83	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010 dominant	612360	67	100	95
NDUFS1	Mitochondrial complex I deficiency, 252010 dominant	157655	76	100	98
NDUFS2	Mitochondrial complex I deficiency, 252010 dominant	602985	89	100	100
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	603846	126	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	602694	81	100	100
NDUFS6	Mitochondrial complex I deficiency, 252010 dominant	603848	65	100	85
NDUFS7	Leigh syndrome, 256000	601825	77	100	100
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	91	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010 dominant	161015	92	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010 dominant	600532	30	68	36
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	45	100	93
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	600644	84	100	100
NEDD4L	Periventricular nodular heterotopia 7, 617201	606384	84	100	97
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	60	95	86
NEXMIF	Mental retardation 98, 300912	300524	44	100	97
NF1	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	80	99	98
NFATC1	No OMIM phenotype	600489	62	100	99
NFIA	Brain malformations and urinary tract defects, 613735	600727	88	100	100
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	90	100	100
NGLY1	Congenital disorder of deglycosylation, 615273	610661	52	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	48	95	92
NID1	No OMIM phenotype	131390	48	99	89
NIN	?Seckel syndrome 7, 614851	608684	89	100	99
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	64	98	93
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550	600635	70	100	97
NLGN3	{Asperger syndrome susceptibility 1}, 300494 {Autism susceptibility 1}, 300425	300336	49	100	97
NLGN4X	{Asperger syndrome susceptibility 2}, 300497 {Autism susceptibility 2}, 300495 Mental retardation, 300495	300427	74	100	100
NLRP3	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900	606416	102	100	100
NONO	Mental retardation, syndromic 34, 300967	300084	27	92	64
NOVA2	No OMIM phenotype	601991	62	91	83
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	83	100	96
NPC2	Niemann-pick disease, type C2, 607625	601015	115	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	78	100	99
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118	600928	51	100	96
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	123	100	100
NRAS	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470	164790	78	100	100
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	102	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NSD1	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	90	100	99
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	57	100	100
NSUN2	Mental retardation 5, 611091	610916	78	100	95
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	75	100	99
NUP62	Striatonigral degeneration, infantile, 271930	605815	67	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	72	100	96
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	79	100	100
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	45	99	93
ODC1	{Colonic adenoma recurrence, reduced risk of}, 114500	165640	88	100	100
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	43	99	88
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	42	100	97
ORC1	Meier-Gorlin syndrome 1, 224690	601902	94	100	100
OTC	Ornithine transcarbamylase deficiency, 311250	300461	59	100	93
P2RX6	No OMIM phenotype	608077	55	87	71
PACS1	Schuurs-Hoeijmakers syndrome, 615009	607492	78	98	95
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	68	100	97
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	112	100	100
PAK3	Mental retardation 30/47, 300558	300142	41	99	86
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	90	100	100
PANX1	No OMIM phenotype	608420	48	100	97
PAX1	?Otofaciocervical syndrome 2, 615560	167411	90	100	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	62	100	99
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	167415	73	100	98
PC	Pyruvate carboxylase deficiency, 266150	608786	84	100	100
PCCA	Propionicacidemia, 606054	232000	55	99	88
PCCB	Propionicacidemia, 606054	232050	87	100	100
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	57	100	97
PCGF2	No OMIM phenotype	600346	46	97	93
PCLO	?Pontocerebellar hypoplasia, type 3, 608027	604918	79	100	99
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	79	100	97
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799	600129	64	99	95
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	64	99	92
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	83	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	65	91	83
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	69	99	94
PEPD	Prolidase deficiency, 170100	613230	68	100	94
PET100	Mitochondrial complex IV deficiency, 220110	614770	35	97	64
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	67	100	98
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	60	95	89
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	92	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	104	100	100
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	91	100	100
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	85	100	95
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	75	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	71	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	94	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	58	100	98
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	93	100	100
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	71	95	91
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	68	100	89
PGAP1	Mental retardation 42, 615802	611655	36	87	68
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	63	100	92
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	42	96	81
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	43	100	93
PHC1	?Microcephaly 11, primary, 615414	602978	73	97	93
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	31	98	73
PHF8	Mental retardation syndrome, Siderius type, 300263	300560	52	99	90
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	86	100	99
PHIP	No OMIM phenotype	612870	76	100	97
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	600286	38	88	76
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	41	93	83
PIGC	No OMIM phenotype	601730	32	88	68
PIGG	Mental retardation 53, 616917	616918	58	99	92
PIGL	CHIME syndrome, 280000	605947	74	100	94
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	66	98	93
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	82	100	100
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	73	100	98
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	102	100	100
PIGW	?Hyperphosphatasia with mental retardation syndrome 5, 616025	610275	62	99	97
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	33	100	68

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	71	100	98
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	70	90	89
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	88	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	76	100	99
PLK4	Microcephaly and chorioretinopathy, 2, 616171	605031	53	92	69
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	71	100	99
PLXNA3	No OMIM phenotype	300022	40	97	89
PLXND1	No OMIM phenotype	604282	55	95	89
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	93	100	100
PMPCA	Spinocerebellar ataxia 2, 213200	613036	43	93	78
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	61	100	98
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	85	100	100
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	90	100	100
POGZ	White-Sutton syndrome, 616364	614787	82	100	97
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	86	100	100
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	88	100	100
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	82	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	82	100	95
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830	614828	85	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	77	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	90	100	100
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	64	98	95
PORCN	Focal dermal hypoplasia, 305600	300651	59	100	96
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	71	100	97
POU3F3	No OMIM phenotype	602480	36	63	50
PPFIA4	No OMIM phenotype	603145	44	98	89
PPM1D	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450	605100	88	100	100
PPOX	Porphyria variegata, 176200	600923	69	100	100
PPP1CB	No OMIM phenotype	600590	38	98	81
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2, 616817	613257	56	99	96
PPP2R1A	Mental retardation 36, 616362	605983	61	100	96
PPP2R5C	No OMIM phenotype	601645	40	87	69
PPP2R5D	Mental retardation 35, 616355	601646	58	100	94
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	81	100	100
PQBP1	Renpenning syndrome, 309500	300463	62	100	100
PRIM1	No OMIM phenotype	176635	36	80	56
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	96	100	100
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157	610087	88	100	98
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	51	95	83

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	59	100	100
PRSS12	Mental retardation 1, 249500	606709	82	100	98
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	98	100	100
PSMD12	No OMIM phenotype	604450	32	83	57
PTCH1	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828	601309	88	99	96
PTCHD1	{Autism, susceptibility to 4}, 300830	300828	54	100	99
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	46	100	94
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	57	77	73
PTF1A	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069	607194	75	95	84
PTGR1	No OMIM phenotype	601274	31	97	74
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	68	99	99
PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263	608625	78	100	98
PTRHD1	No OMIM phenotype	617342	63	100	93
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	58	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PUF60	Verheij syndrome, 615583	604819	71	97	95
PURA	Mental retardation 31, 616158	600473	87	98	86
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	71	100	98
PUS3	?Mental retardation 55, 617051	616283	62	100	99
PUS7	No OMIM phenotype	616261	56	97	90
PYCR1	Cutis laxa, type IIB, 612940 Cutis laxa, type IIIB, 614438	179035	71	100	96
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	60	96	87
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	57	99	92
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	60	100	95
RAB18	Warburg micro syndrome 3, 614222	602207	59	97	91
RAB27A	Griscelli syndrome, type 2, 607624	603868	83	100	100
RAB39B	Mental retardation 72, 300271 ?Waisman syndrome, 311510	300774	48	100	97
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	68	100	98
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	69	100	96
RAB40AL	No OMIM phenotype	300405	50	100	100
RAC1	No OMIM phenotype	602048	43	88	69
RAD21	Cornelia de Lange syndrome 4, 614701	606462	65	100	93
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	66	97	92
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	82	100	98
RAI1	Smith-Magenis syndrome, 182290	607642	91	100	100
RARB	Microphthalmia, syndromic 12, 615524	180220	44	100	94
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	71	100	95
RBBP8	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744	604124	64	100	94
RBFOX1	No OMIM phenotype	605104	75	100	97
RBM10	TARP syndrome, 311900	300080	42	98	88
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	87	100	100
RBPJ	Adams-Oliver syndrome 3, 614814	147183	61	92	83
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175	607867	88	100	100
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	91	100	98
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975	605226	36	93	76

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
REV3L	No OMIM phenotype	602776	55	97	88
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	62	100	98
RHEB	No OMIM phenotype	601293	14	59	24
RIT1	Noonan syndrome 8, 615355	609591	54	100	98
RLIM	Mental retardation 61, 300978	300379	35	98	84
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	82	100	100
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	89	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	50	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	112	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	54	100	95
RNF113A	?Trichothiodystrophy 5, nonphotosensitive, 300953	300951	58	100	100
RNF125	Tenorio syndrome, 616260	610432	56	100	86
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	57	100	97
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	63	100	98
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	77	97	95
RPL10	{Autism, susceptibility to 5}, 300847	312173	68	100	100
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation 19, 300844	300075	40	96	81
RSPRY1	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723	616585	54	100	93
RTEL1	Dyskeratosis congenita 4, 615190 Dyskeratosis congenita 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	58	97	87
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833	610436	71	99	96
RUBCN	?Spinocerebellar ataxia 15, 615705	613516	76	98	96
RUSC2	No OMIM phenotype	611053	81	100	99
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480	602218	99	100	98
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	76	100	94
SATB2	Glass syndrome, 612313	608148	67	100	99
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	61	100	96
SC5D	Lathosterolosis, 607330	602286	86	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SCAF4	No OMIM phenotype	616023	39	92	72
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	100	100	99
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	102	97	95
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	100	100	99
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	97	100	99
SCO1	Mitochondrial complex IV deficiency, 220110	603644	69	100	98
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	94	100	100
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	43	77	64
SEMA3E	?CHARGE syndrome, 214800	608166	72	100	100
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	92	100	100
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	69	100	99
SETBP1	Mental retardation 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	74	98	97
SETD1A	No OMIM phenotype	611052	57	97	91
SETD2	Luscan-Lumish syndrome, 616831	612778	75	100	98
SETD5	Mental retardation 23, 615761	615743	57	99	96
SF1	No OMIM phenotype	601516	36	83	69
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	86	94	94
SH3KBP1	No OMIM phenotype	300374	25	88	62
SHANK2	{Autism susceptibility 17}, 613436	603290	81	100	100
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	56	93	82

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	80	100	97
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	78	100	100
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434	300579	44	100	93
SIK1	Epileptic encephalopathy, early infantile, 30, 616341	605705	52	96	86
SIL1	Marinesco-Sjogren syndrome, 248800	608005	85	100	100
SIN3A	Witteveen-Kolk syndrome, 613406	607776	42	96	83
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	103	100	100
SKI	Shprintzen-Goldberg syndrome, 182212	164780	55	99	97
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	71	100	99
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905	608305	55	100	94
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	42	99	89
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	75	100	99
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	118	100	100
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	95	100	100
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105	600300	38	98	78
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	600229	62	99	90
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	87	100	100
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	106	90	86
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	85	100	100
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	63	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	104	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	62	100	96
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	76	100	100
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	29	98	80
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	30	94	67

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	103	100	100
SLC39A12	No OMIM phenotype	608734	39	97	81
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721	608732	48	100	93
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	71	100	99
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	86	100	100
SLC6A17	Mental retardation 48, 616269	610299	69	100	98
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	87	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	19	81	42
SLC7A7	Lysinuric protein intolerance, 222700	603593	79	100	98
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	50	100	96
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	76	100	98
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	78	98	96
SMARCA4	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325	603254	91	100	100
SMARCB1	Coffin-Siris syndrome 3, 614608 {Rhabdoid predisposition syndrome 1}, 609322 Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091	601607	123	100	100
SMARCC2	No OMIM phenotype	601734	42	96	83
SMARCE1	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174	603111	48	86	75
SMC1A	Cornelia de Lange syndrome 2, 300590	300040	48	100	91
SMC3	Cornelia de Lange syndrome 3, 610759	606062	50	93	84
SMOC1	Microphthalmia with limb anomalies, 206920	608488	77	100	100
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	110	100	100
SMS	Mental retardation, Snyder-Robinson type, 309583	300105	39	95	94
SNAP25	?Myasthenic syndrome, congenital, 18, 616330	600322	81	100	100
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	99	100	100
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	88	100	100
SNRNP70	No OMIM phenotype	180740	49	99	94
SNRPN	Prader-Willi syndrome, 176270	182279	90	100	98
SNX14	Spinocerebellar ataxia 20, 616354	616105	27	80	52
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	79	98	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SON	ZTTK syndrome, 617140	182465	55	91	80
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	65	100	95
SOS2	Noonan syndrome 9, 616559	601247	34	93	70
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	45	99	87
SOX11	Mental retardation, 27, 615866	600898	64	100	96
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	113	100	100
SOX3	Mental retardation, with isolated growth hormone deficiency, 300123 Panhypopituitarism, 312000	313430	34	99	76
SOX5	Lamb-Shaffer syndrome, 616803	604975	72	100	100
SPAST	Spastic paraplegia 4, 182601	604277	49	100	91
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577	613940	46	99	88
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	73	100	99
SPOCK1	No OMIM phenotype	602264	43	99	85
SPRED1	Legius syndrome, 611431	609291	67	100	95
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	87	100	100
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia 14, 615386	604985	82	100	99
SRCAP	Floating-Harbor syndrome, 136140	611421	101	100	99
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	105	100	100
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	50	99	91
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	37	100	92
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation 12, 611090	606494	111	100	100
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	85	93	93
STAG1	No OMIM phenotype	604358	39	92	74
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	37	96	79
STIL	Microcephaly 7, primary, 612703	181590	81	100	97
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	58	100	100
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	83	100	100
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	77	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	82	100	99
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172	601485	57	99	87
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	92	100	100
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	38	86	75
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	74	100	98
SUCO	No OMIM phenotype	No id	57	99	92
SUMF1	Multiple sulfatase deficiency, 272200	607939	87	100	100
SUOX	Sulfite oxidase deficiency, 272300	606887	104	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	71	88	88
SUZ12	No OMIM phenotype	606245	54	98	83
SYN1	Epilepsy, with variable learning disabilities and behavior disorders, 300491 dominant	313440	37	86	67
SYNCRIP	No OMIM phenotype	616686	30	79	54
SYNE1	Emery-Dreifuss muscular dystrophy 4, 612998 Spinocerebellar ataxia 8, 610743	608441	87	100	100
SYNGAP1	Mental retardation 5, 612621	603384	55	95	82
SYP	Mental retardation 96, 300802	313475	48	100	100
SYT1	No OMIM phenotype	185605	52	96	88
SYT14	Spinocerebellar ataxia 11, 614229	610949	92	93	89
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	57	99	93
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	78	100	100
TAF2	Mental retardation 40, 615599	604912	63	100	99
TAT	Tyrosinemia, type II, 276600	613018	82	100	100
TBC1D20	Warburg micro syndrome 4, 615663	611663	51	93	85
TBC1D24	DOOR syndrome, 220500 Deafness 86, 614617 Deafness 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	92	100	100
TBC1D7	Macrocephaly/megalencephaly syndrome, 248000	612655	39	95	85
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	604649	58	95	87
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	80	99	91
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	616899	35	89	72

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TBL1XR1	Mental retardation 41, 616944 Pierpont syndrome, 602342	608628	47	91	83
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	96	100	100
TBR1	No OMIM phenotype	604616	62	100	95
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430	602054	55	79	66
TCF12	Craniosynostosis 3, 615314	600480	51	100	94
TCF20	No OMIM phenotype	603107	60	100	99
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954	602272	73	100	98
TCF7L2	{Diabetes mellitus, type 2, susceptibility to}, 125853	602228	84	100	99
TCN2	Transcobalamin II deficiency, 275350	613441	110	100	100
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	60	100	96
TDP2	Spinocerebellar ataxia 23, 616949	605764	73	100	91
TECPR2	Spastic paraplegia 49, 615031	615000	92	100	100
TECR	Mental retardation 14, 614020	610057	68	99	94
TELO2	You-Hoover-Fong syndrome, 616954	611140	57	98	92
TFAP2A	Branchiooculofacial syndrome, 113620	107580	66	100	96
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	111	94	94
TGFBR2	No OMIM phenotype	190182	100	100	100
TGIF1	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	602630	97	100	100
TH	Holoprosencephaly 4, 142946	191290	55	100	95
THOC2	Segawa syndrome, recessive, 605407	300395	38	98	87
THOC6	Mental retardation 12/35, 300957	615403	101	100	100
THRB	Beaulieu-Boycott-Innes syndrome, 613680	190160	89	100	100
TIMM8A	Thyroid hormone resistance, 188570 Thyroid hormone resistance, 274300 Thyroid hormone resistance, selective pituitary, 145650	300356	42	100	100
TINF2	Mohr-Tranebjaerg syndrome, 304700	604319	92	100	100
TLK2	Dyskeratosis congenita 3, 613990 Revesz syndrome, 268130	608439	31	88	67

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TM4SF20	No OMIM phenotype	615404	61	100	100
TMCO1	{Specific language impairment 5}, 615432	614123	60	100	98
TMEM165	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614726	76	100	100
TMEM231	Congenital disorder of glycosylation, type IIk, 614727	614949	56	98	93
TMEM237	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614423	80	100	99
TMEM240	Joubert syndrome 14, 614424	616101	62	100	100
TMEM5	Spinocerebellar ataxia 21, 607454	605862	106	100	99
TMEM67	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	609884	51	99	90
TMEM70	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	612418	75	100	99
TMLHE	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	300777	42	98	87
TMPRSS7	{Autism, susceptibility to 6}, 300872	617218	47	100	92
TMTC3	Lissencephaly 8, 617255	617218	59	100	95
TMX2	No OMIM phenotype	616715	46	96	85
TNIK	Mental retardation 54, 617028	610005	43	98	84
TOE1	Pontocerebellar hypoplasia, type 7, 614969	613931	58	100	98
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	41	92	81
TPO	Thyroid dyshormonogenesis 2A, 274500	606765	84	99	92
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	79	100	98
TRAIP	Seckel syndrome 9, 616777	605958	48	100	94
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	44	96	82
TRAPPC9	Mental retardation 13, 613192	611966	85	100	99
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	129	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	77	100	100
TRIO	Mental retardation 44, 617061	601893	86	99	98
TRIP12	No OMIM phenotype	604506	75	100	99
TRMT1	No OMIM phenotype	611669	54	97	86
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	616013	46	99	83

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	94	100	99
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	94	100	100
TSEN15	Pontocerebellar hypoplasia, type 2F, 617026	608756	35	88	74
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	96	96
TSHB	Hypothryoidism, congenital, nongoitrous 4, 275100	188540	120	100	100
TSPAN7	Mental retardation 58, 300210	300096	40	100	85
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	57	90	78
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	77	100	98
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	64	100	90
TTI2	Mental retardation 39, 615541	614426	79	100	100
TUBA1A	Lissencephaly 3, 611603	602529	34	93	78
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180	605742	116	100	99
TUBB	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610	191130	55	90	83
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	51	100	89
TUBB2B	Polymicrogyria, symmetric or asymmetric, 610031	612850	65	100	94
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	75	100	100
TUBB4A	Dystonia 4, torsion, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	60	97	93
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	65	100	99
TUBGCP4	Microcephaly and chorioretinopathy, 3, 616335	609610	42	96	82
TUBGCP6	Microcephaly and chorioretinopathy, 1, 251270	610053	94	100	100
TUSC3	Mental retardation 7, 611093	601385	74	100	100
TWIST1	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400	601622	79	100	86
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	103	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
UBA5	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia 24, 617133	610552	31	79	51
UBE2A	Mental retardation syndromic, Nascimento-type, 300860	312180	33	100	96
UBE3A	Angelman syndrome, 105830	601623	61	100	99
UBE3B	Kaufman oculocerebrofacial syndrome, 244450	608047	49	99	90
UBR1	Johanson-Blizzard syndrome, 243800	605981	69	100	95
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801	612636	89	100	99
UPB1	Beta-ureidopropionase deficiency, 613161	606673	108	100	100
UPF3B	Mental retardation, syndromic 14, 300676	300298	32	99	83
UQCC2	?Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	51	100	91
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	167	100	100
UROC1	?Urocanase deficiency, 276880	613012	82	100	100
USP18	Pseudo-TORCH syndrome 2, 617397	607057	58	96	94
USP27X	Mental retardation 105, 300984	300975	50	100	98
USP7	No OMIM phenotype	602519	33	85	63
USP9X	Mental retardation 99, 300919 Mental retardation 99, syndromic, female-restricted, 300968	300072	54	100	96
UTRN	No OMIM phenotype	128240	82	100	97
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	105	100	99
VPS11	Leukodystrophy, hypomyelinating, 12, 616683	608549	51	99	89
VPS13B	Cohen syndrome, 216550	607817	83	100	99
VPS13C	Parkinson disease 23, early onset, 616840	608879	37	90	72
VPS37A	Spastic paraplegia 53, 614898	609927	46	76	55
VPS53	Pontocerebellar hypoplasia, type 2E, 615851	615850	49	98	91
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	78	100	96
VWA3B	?Spinocerebellar ataxia 22, 616948	614884	48	98	90
WAC	Desanto-Shinawi syndrome, 616708	615049	48	97	84
WASHC4	?Mental retardation 43, 615817	615748	70	99	95
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	96	100	99
WDR13	No OMIM phenotype	300512	56	100	98
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	73	100	98
WDR4	No OMIM phenotype	605924	79	100	99
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	32	89	78
WDR54	No OMIM phenotype	No id	45	96	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
WDR62	Microcephaly 2, primary, with or without cortical malformations, 604317	613583	90	100	100
WDR73	Galloway-Mowat syndrome, 251300	616144	58	99	91
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185	614218	90	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	102	100	100
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia 12, 614322	605131	72	100	100
XPA	Xeroderma pigmentosum, group A, 278700	611153	47	98	86
XPC	Xeroderma pigmentosum, group C, 278720	613208	95	100	100
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	85	100	99
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	75	94	88
YAP1	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	40	86	73
YME1L1	?Optic atrophy 11, 617302	607472	36	90	71
YWHAE	No OMIM phenotype	605066	56	99	80
YY1	No OMIM phenotype	600013	82	100	95
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	79	100	100
ZBTB18	Mental retardation 22, 612337	608433	103	100	100
ZBTB20	Primrose syndrome, 259050	606025	91	100	99
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069	614064	82	100	100
ZC3H14	Mental retardation 56, 617125	613279	96	100	100
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	25	97	70
ZDHHC15	?Mental retardation 91, 300577	300576	38	100	94
ZDHHC9	Mental retardation syndromic, Raymond type, 300799	300646	44	100	90
ZEB2	Mowat-Wilson syndrome, 235730	605802	81	100	100
ZFYVE26	Spastic paraplegia 15, 270700	612012	79	100	97
ZIC1	Craniosynostosis 6, 616602	600470	108	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	92	93	88
ZIC4	No OMIM phenotype	608948	104	100	100
ZMYND11	Mental retardation 30, 616083	608668	43	98	88
ZNF101	No OMIM phenotype	603983	33	95	83
ZNF292	No OMIM phenotype	616213	54	97	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ZNF407	No OMIM phenotype	615894	56	98	94
ZNF41	No OMIM phenotype	314995	40	100	98
ZNF592	No OMIM phenotype	613624	80	100	99
ZNF674	No OMIM phenotype	300573	53	100	100
ZNF711	Mental retardation 97, 300803	314990	39	100	92
ZNF81	No OMIM phenotype	314998	36	100	92
ZSWIM6	Acromelic frontonasal dysostosis, 603671	615951	51	94	87

- 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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x