Whole Exome Sequencing
Gene package Intellectual disability, version 4, 5-8-2016

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
ABCC9	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 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, X-linked 63, 300387	300157	44	99	91
ACTB	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310	102630	77	100	100
ACTG1	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583	102560	104	100	100
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	86	100	100
ACY1	Aminoacylase 1 deficiency, 609924	104620	84	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ADAR	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010	146920	96	100	100
ADAT3	Mental retardation, autosomal recessive 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, X-linked, FRAXE type, 309548	300806	45	100	93
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
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, X-linked 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
AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Ovarian cancer, somatic, 167000 {Schizophrenia, susceptibility to}, 181500 Proteus syndrome, somatic, 176920 Cowden syndrome 6, 615109	164730	87	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	33	89	66
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603	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
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	101	100	100
ALG13	Epileptic encephalopathy, early infantile, 36, 300884	300776	30	88	60

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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
ALG9	Congenital disorder of glycosylation, type Il, 608776	606941	68	100	97
ALX1	?Frontonasal dysplasia 3, 613456	601527	96	100	100
ALX4	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451 {Craniosynostosis 5, susceptibility to}, 615529	605420	82	100	100
AMPD2	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809	102771	60	100	94
AMT	Glycine encephalopathy, 605899	238310	91	100	100
ANK3	?Mental retardation, autosomal recessive, 37, 615493	600465	72	100	99
ANKEF1	No OMIM phenotype	NA	53	99	95
ANKH	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600	605145	79	100	100
ANKLE2	?Microcephaly 16, primary, autosomal recessive, 616681	616062	64	98	90
ANKRD11	KBG syndrome, 148050	611192	77	100	97
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728	613726	73	100	100
ANTXR1	{Hemangioma, capillary infantile, susceptibility to}, 602089 GAPO syndrome, 230740	606410	78	98	91
AP1S2	Mental retardation, X-linked syndromic 5, 304340	300629	32	97	79
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	73	100	95
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066	607245	83	100	100
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744	607244	61	100	98
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936	602296	75	100	100
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067	607243	40	97	85
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
ARHGEF6	Mental retardation, X-linked 46, 300436	300267	50	99	88
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	47	100	87
ARID1A	Mental retardation, autosomal dominant 14, 614607	603024	83	97	94
ARID1B	Mental retardation, autosomal dominant 12, 614562	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 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900 ?Retinitis pigmentosa 55, 613575	608845	48	95	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950	300180	44	94	77
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215	300382	24	84	61
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, autosomal recessive, 608716	605481	62	100	96
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	77	100	98
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	53	99	94
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	71	100	99
ATP1A2	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481	182340	109	100	100
ATP1A3	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338	182350	96	100	99
ATP2A2	Darier disease, 124200 Acrokeratosis verruciformis, 101900	108740	95	100	100
ATP6AP2	?Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911	300556	39	100	90
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	94	100	100
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489	300011	53	99	94
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	82	100	98
ATR	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564	601215	81	100	98
ATRIP	No OMIM phenotype	606605	59	99	91
ATRX	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580	300032	38	98	83
AUH	3-methylglutaconic aciduria, type I, 250950	600529	60	100	90
AUTS2	Mental retardation, autosomal dominant 26, 615834	607270	81	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	83	100	97
B3GLCT	Peters-plus syndrome, 261540	610308	65	98	90
B3GNT2	No OMIM phenotype	605581	108	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	70	100	100
B4GALT7	Ehlers-Danlos syndrome, progeroid type, 1, 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
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	No OMIM phenotype	606557	73	100	100
BCOR	Microphthalmia, syndromic 2, 300166	300485	52	99	94
BCS1L	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358	603647	114	100	100
BLM	Bloom syndrome, 210900	604610	74	100	99
BRAF	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707	164757	60	98	88
BRPF1	No OMIM phenotype	602410	70	100	97
BRWD3	Mental retardation, X-linked 93, 300659	300553	48	98	90
BSCL2	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924	606158	82	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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, autosomal recessive, 615035	613541	56	100	100
C1orf167	No OMIM phenotype	NA	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, autosomal recessive 3, with renal tubular acidosis, 259730	611492	89	100	100
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	66	100	100
CACNG2	Mental retardation, autosomal dominant 10, 614256	602911	100	100	100
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	100	100	99
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422	300172	39	98	87
CBL	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, autosomal recessive 3, 608443	610055	74	100	99
CC2D2A	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360	612013	82	100	99
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	82	100	96
CCDC13	No OMIM phenotype	NA	45	99	89
CCDC78	Myopathy, centronuclear, 4, 614807	614666	64	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, autosomal dominant 3, 612580	114019	84	100	97
CDK5	?Lissencephaly 7 with cerebellar hypoplasia, 616342	123831	45	100	87
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804	608201	69	100	97
CDK6	?Microcephaly 12, primary, autosomal recessive, 616080	603368	77	99	95
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	47	98	94
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	46	80	72
CDON	Holoprosencephaly 11, 614226	608707	90	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CECR1	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	34	93	73
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676	609279	80	100	98
CEP104	Joubert syndrome 25, 616781	616690	47	99	88
CEP135	?Microcephaly 8, primary, autosomal recessive, 614673	611423	60	100	97
CEP152	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823	613529	76	100	98
CEP290	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 ?Bardet-Biedl syndrome 14, 615991	610142	55	98	92
CEP41	Joubert syndrome 15, 614464	610523	64	100	93
CEP63	?Seckel syndrome 6, 614728	614724	69	100	94
CHAMP1	Mental retardation, autosomal dominant 40, 616579	616327	80	100	100
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	75	100	98
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
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	70	98	86
CLIC2	?Mental retardation, X-linked, syndromic 32, 300886	300138	46	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	68	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	73	100	94
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
CNKSR2	No OMIM phenotype	300724	42	98	90
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042	604569	85	100	100
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	91	100	98
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	72	99	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	79	100	100
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	72	100	100
COL18A1	Knobloch syndrome, type 1, 267750	120328	71	100	95
COL4A1	Porencephaly 1, 175780 Brain small vessel disease with or without ocular anomalies, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000	120130	76	99	96
COL4A2	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519	120090	69	100	97
COL4A3BP	Mental retardation, autosomal dominant 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	No coverage data		
COX10	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000	602125	105	100	100
COX15	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119	603646	86	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, autosomal recessive 34, 614499	603454	73	100	96
CRB2	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	609720	57	98	89
CRBN	Mental retardation, autosomal recessive 2, 607417	609262	87	100	94
CREBBP	Rubinstein-Taybi syndrome, 180849	600140	81	100	97
CSNK2A1	No OMIM phenotype	115440	43	95	75
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	71	100	100
CTCF	Mental retardation, autosomal dominant 21, 615502	604167	47	96	89
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	74	95	88
CTNNB1	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Pilomatricoma, somatic, 132600 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550	116806	81	100	100
CTNND1	No OMIM phenotype	601045	49	99	92
CTNND2	No OMIM phenotype	604275	76	94	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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, X-linked, syndromic 15 (Cabezas type), 300354	300304	39	100	92
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	86	100	100
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	91	100	100
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538	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
DCHS1	Van Maldergem syndrome 1, 601390 Mitral valve prolapse 2, 607829	603057	70	99	97
DCPS	Al-Raqad syndrome, 616459	610534	61	100	94
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067	300121	47	98	85
DDHD2	Spastic paraplegia 54, autosomal recessive, 615033	615003	86	100	100
DDX11	Warsaw breakage syndrome, 613398	601150	39	78	66
DDX3X	Mental retardation, X-linked 102, 300958	300160	50	99	94
DEAF1	Mental retardation, autosomal dominant 24, 615828	602635	81	87	82
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, autosomal dominant 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, X-linked, 305000	300126	57	100	94
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	84	100	97
DLG3	Mental retardation, X-linked 90, 300850	300189	40	100	84
DLG4	No OMIM phenotype	602887	65	100	96
DMD	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045	300377	50	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DMPK	Myotonic dystrophy 1, 160900	605377	70	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
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700	611432	84	100	99
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	80	100	99
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	69	100	100
DPP6	{Ventricular fibrillation, paroxysmal familial, 2}, 612956 Mental retardation, autosomal dominant 33, 616311	126141	70	99	94
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	79	98	95
DST	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425	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, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	86	100	99
DYRK1A	Mental retardation, autosomal dominant 7, 614104	600855	99	100	100
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960	300205	43	100	93
EDC3	?Mental retardation, autosomal recessive 50, 616460	609842	46	99	89
EDNRB	{Hirschsprung disease, susceptibility to, 2}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580	131244	93	100	100
EDRF1	No OMIM phenotype	NA	71	100	99
EEF1A2	Mental retardation, autosomal dominant 38, 616393 Epileptic encephalopathy, early infantile, 33, 616409	602959	78	100	96
EFCAB1	No OMIM phenotype	NA	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
EIF4G1	{Parkinson disease 18}, 614251	600495	92	100	99
ELOVL4	Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190	605512	57	100	98
EMG1	Bowen-Conradi syndrome, 211180	611531	78	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
EML1	No OMIM phenotype	602033	46	99	89
EMX2	Schizencephaly, 269160	600035	95	100	100
EOMES	No OMIM phenotype	604615	54	100	87
EP300	Rubinstein-Taybi syndrome 2, 613684 Colorectal cancer, somatic, 114500	602700	90	100	100
EPB41L1	?Mental retardation, autosomal dominant 11, 614257	602879	88	100	99
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	73	100	92
ERCC2	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 Cerebrooculofacioskeletal syndrome 2, 610756	126340	77	100	100
ERCC3	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390	133510	81	100	100
ERCC5	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Cerebrooculofacioskeletal syndrome 3, 616570	133530	73	100	99
ERCC6	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980	609413	93	100	100
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	60	100	96
ERLIN2	Spastic paraplegia 18, autosomal recessive, 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
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	81	100	100
EZH2	Weaver syndrome, 277590	601573	84	100	99
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	67	100	94
FAT4	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006	612411	78	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FBN1	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370 Geleophysic dysplasia 2, 614185	134797	86	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	90	100	98
FBXO31	?Mental retardation, autosomal recessive 45, 615979	609102	42	94	83
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	43	99	87
FGFR1	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465	136350	99	100	100
FGFR2	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Scaphocephaly and Axenfeld-Rieger anomaly LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592	176943	80	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FGFR3	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500 Cervical cancer, somatic, 603956 LADD syndrome, 149730 CATSHL syndrome, 610474 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Spermatocytic seminoma, somatic, 273300 SADDAN, 616482	134934	73	100	100
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	84	97	91
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	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	75	100	100
FLNA	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048	300017	55	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	82	100	97
FMN2	Mental retardation, autosomal recessive 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
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	616305	51	99	89
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	70	100	100
FTSJ1	Mental retardation, X-linked 9, 309549	300499	59	100	100
FUCA1	Fucosidosis, 230000	612280	96	100	100
GABRA1	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	95	100	100
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, autosomal dominant 18, 615074	614998	40	98	82
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	93	100	100
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, X-linked 41, 300849	300104	64	100	98
GFAP	Alexander disease, 203450	137780	62	100	100
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800	304040	54	100	100
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480	608803	40	83	58
GK	Glycerol kinase deficiency, 307030	300474	33	99	74
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GLI2	Holoprosencephaly-9, 610829 Culler-Jones syndrome, 615849	165230	82	99	97
GLI3	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200 {Hypothalamic hamartomas, somatic}, 241800	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 (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351	615320	100	100	100
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	66	100	99
GNAQ	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000	600998	35	69	47
GNAS	Pseudohypoparathyroidism Ia, 103580 McCune-Albright syndrome, somatic, mosaic 174800 Pseudohypoparathyroidism Ic, 612462 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 Acromegaly, somatic, 102200 Pseudopseudohypoparathyroidism, 612463 ACTH-independent macronodular adrenal hyperplasia, 219080	139320	91	100	97
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	92	100	100
GNS	Mucopolysaccharidosis type IIID, 252940	607664	84	100	93
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, autosomal recessive 49, 616281	138210	55	99	91
GRIA3	Mental retardation, X-linked 94, 300699	305915	54	100	98
GRID2	Spinocerebellar ataxia, autosomal recessive 18, 616204	602368	93	100	100
GRIK2	Mental retardation, autosomal recessive, 6, 611092	138244	88	100	99
GRIN1	Mental retardation, autosomal dominant 8, 614254	138249	96	100	99
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	80	100	100
GRIN2B	Mental retardation, autosomal dominant 6, 613970 Epileptic encephalopathy, early infantile, 27, 616139	138252	95	100	100
GRIN3B	No OMIM phenotype	606651	59	85	75

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GRM1	Spinocerebellar ataxia, autosomal recessive 13, 614831	604473	89	100	100
GSE1	No OMIM phenotype	616886	68	100	97
GSS	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130	601002	80	100	99
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	112	100	100
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, autosomal recessive, 610738	605998	100	100	100
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	53	100	99
HCFC1	Mental retardation, X-linked 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	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882	300269	41	100	94
HECTD1	No OMIM phenotype	NA	52	96	88
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	605109	54	99	92
HERC2	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 Mental retardation, autosomal recessive 38, 615516	605837	37	73	63
HESX1	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230	601802	78	100	100
HEXA	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800	606869	85	100	100
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	88	100	95
HIVEP2	No OMIM phenotype	143054	69	100	99
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	88	100	100
HNMT	{Asthma, susceptibility to}, 600807 Mental retardation, autosomal recessive 51, 616739	605238	63	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HOXA1	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536	142955	108	100	100
HPD	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350	609695	102	100	100
HPRT1	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323	308000	36	100	85
HRAS	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Spitz nevus or nevus spilus, somatic}, 137550	190020	128	100	100
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220	300256	63	100	100
HSPD1	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233	118190	27	84	58
HUWE1	Mental retardation, X-linked syndromic, Turner type, 300706	300697	43	99	89
IBA57	?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451	615316	59	95	90
IDS	Mucopolysaccharidosis II, 309900	300823	62	100	100
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015	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	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Retinitis pigmentosa 71, 616394	607386	77	100	99
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	Incontinentia pigmenti, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 Immunodeficiency 33, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	25	75	49
IL1RAPL1	Mental retardation, X-linked 21/34, 300143	300206	50	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
INPP5B	No OMIM phenotype	147264	86	100	99
INPP5E	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300	613037	71	98	92
INTS1	No OMIM phenotype	611345	50	98	89
INTS8	No OMIM phenotype	611351	40	92	70
IQSEC2	Mental retardation, X-linked 1, 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
ITPR1	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
JAG1	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon	601920	89	99	96
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	102	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, autosomal dominant 32, 616268	601408	82	100	98
KAT6B	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170	605880	94	100	99
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	67	100	98
KCNC3	Spinocerebellar ataxia 13, 605259	176264	56	80	66
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500	603305	65	99	95
KCNJ10	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791	602208	122	100	100
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582 Maturity-onset diabetes of the young, type 13, 616329	600937	104	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		

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KCNQ2	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720	602235	70	100	100
KCNQ5	No OMIM phenotype	607357	76	98	95
KCNT1	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005	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
KDM5C	Mental retardation, X-linked, 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
KIAA0196	Spastic paraplegia 8, autosomal dominant, 603563 Ritscher-Schinzel syndrome 1, 220210	610657	96	100	99
KIAA1033	?Mental retardation, autosomal recessive 43, 615817	615748	70	99	95
KIAA1109	No OMIM phenotype	611565	49	96	86
KIAA1586	No OMIM phenotype	NA	31	92	66
KIAA2022	Mental retardation, X-linked 98, 300912	300524	44	100	97
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	53	99	95
KIF1A	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255	601255	75	100	98
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	No coverage data		
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	36	87	63
KIF4A	?Mental retardation, X-linked 100, 300923	300521	23	87	57
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282	604593	42	96	79
KIF7	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131	611254	64	98	93
KIRREL3	Mental retardation, autosomal dominant 4, 612581	607761	77	100	99
KMT2A	Wiedemann-Steiner syndrome, 605130 Leukemia, myeloid/lymphoid or mixed-lineage	159555	55	99	95
KMT2D	Kabuki syndrome 1, 147920	602113	80	100	99
KNL1	Microcephaly 4, primary, autosomal recessive, 604321	609173	No coverage data		
KPTN	Mental retardation, autosomal recessive 41, 615637	615620	54	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KRAS	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Breast cancer, somatic, 114480 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 RAS-associated autoimmune leukoproliferative disorder, 614470	190070	64	100	100
KRBOX4	No OMIM phenotype	300585	59	100	100
L1CAM	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100	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
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	76	100	100
LINS1	Mental retardation, autosomal recessive 27, 614340	610350	No coverage data		
LRP2	Donnai-Barrow syndrome, 222448	600073	89	100	100
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	76	100	98
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	96	100	100
MAGEC3	No OMIM phenotype	300469	25	91	66
MAGEL2	Schaaf-Yang syndrome, 615547	605283	70	100	97
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	44	100	89
MAN1B1	Mental retardation, autosomal recessive 15, 614202	604346	91	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	86	100	98
MANBA	Mannosidosis, beta, 248510	609489	84	100	100
MAOA	Brunner syndrome, 300615	309850	39	100	86
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	87	100	100
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	91	100	100
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850	610550	89	100	100
MBD5	Mental retardation, autosomal dominant 1, 156200	611472	82	100	100
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 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
MCOLN1	Mucolipidosis IV, 252650	605248	95	100	99
MCOLN3	No OMIM phenotype	607400	37	94	78
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200	607117	67	100	99
MECP2	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Mental retardation, X-linked syndromic, Lubs type, 300260 Rett syndrome, atypical, 312750	300005	52	99	81
MED12	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895	300188	47	100	91
MED13L	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789	608771	77	100	99
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	77	100	100
MED23	Mental retardation, autosomal recessive 18, 614249	605042	72	100	97
MEF2C	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443	600662	66	100	98
METTL23	Mental retardation, autosomal recessive 44, 615942	615262	42	100	100
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	87	100	100
MID1	Opitz GBBB syndrome, type I, 300000	300552	57	100	99
MID2	?Mental retardation, X-linked 101, 300928	300204	33	96	76
MKKS	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231	604896	106	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 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, autosomal recessive 2, 615219	603785	76	100	97
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	77	100	100
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	75	100	99
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050	607093	94	100	100
MTMR8	No OMIM phenotype	NA	24	97	66
MTOR	Smith-Kingsmore syndrome, 616638	601231	47	98	86
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	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900	251170	94	100	100
MYCN	Feingold syndrome, 164280	164840	68	100	91
MYH9	May-Hegglin anomaly, 155100 Fechtner syndrome, 153640 Sebastian syndrome, 605249 Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Macrothrombocytopenia and progressive sensorineural deafness, 600208	160775	91	100	100
MYO5A	Griscelli syndrome, type 1, 214450	160777	74	100	98
MYT1L	Mental retardation, autosomal dominant 39, 616521	613084	89	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NAA10	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800	300013	51	100	100
NAA15	No OMIM phenotype	608000	32	91	68
NAGA	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241	104170	86	100	100
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491	609701	81	100	93
NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266	611549	43	98	87
NANS	No OMIM phenotype	605202	36	99	87
NBN	Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065	602667	66	100	97
NCOR2	No OMIM phenotype	600848	42	95	80
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	94	100	100
NDP	Norrie disease, 310600 Exudative vitreoretinopathy 2, X-linked, 305390	300658	68	100	100
NDST1	Mental retardation, autosomal recessive 46, 616116	600853	109	100	100
NDUFA1	Mitochondrial complex I deficiency, 252010	300078	85	100	90
NDUFA11	Mitochondrial complex I deficiency, 252010	612638	57	100	96
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000	614530	98	100	100
NDUFS1	Mitochondrial complex I deficiency, 252010	157655	76	100	98
NDUFS2	Mitochondrial complex I deficiency, 252010	602985	89	100	100
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	603846	126	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010	602694	81	100	100
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	161015	92	100	100
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	45	100	93
NEDD4L	No OMIM phenotype	606384	84	100	97
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	60	95	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NF1	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	80	99	98
NFATC1	No OMIM phenotype	600489	62	100	99
NFIA	No OMIM phenotype	600727	88	100	100
NFIX	Sotos syndrome 2, 614753 Marshall-Smith syndrome, 602535	164005	90	100	100
NHS	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200	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	{Thyroid cancer, monmedullary, 1}, 188550 Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978	600635	70	100	97
NLGN3	{Autism susceptibility, X-linked 1}, 300425 {Asperger syndrome susceptibility, X-linked 1}, 300494	300336	49	100	97
NLGN4X	{Autism susceptibility, X-linked 2}, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 Mental retardation, X-linked, 300495	300427	74	100	100
NLRP3	Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115	606416	102	100	100
NPHP1	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583	607100	78	100	99
NPRL3	No OMIM phenotype	600928	51	100	96
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	123	100	100
NRAS	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	164790	78	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	102	100	99
NSD1	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650	606681	90	100	99
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	57	100	100
NSUN2	Mental retardation, autosomal recessive 5, 611091	610916	78	100	95
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	75	100	99
OCLN	Band-like calcification with simplified gyration and polymicrogyria, 251290	602876	79	100	100
OCRL	Lowe syndrome, 309000 Dent disease 2, 300555	300535	45	99	93
ODC1	{Colonic adenoma recurrence, reduced risk of}, 114500	165640	88	100	100
OFD1	Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804 ?Retinitis pigmentosa 23, 300424	300170	43	99	88
OPHN1	Mental retardation, X-linked, 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	Mental retardation, autosomal dominant 17, 615009	607492	78	98	95
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	68	100	97
PAH	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600	612349	112	100	100
PAK3	Mental retardation, X-linked 30/47, 300558	300142	41	99	86
PANK2	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236	606157	90	100	100
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 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hypoplasia 1, 136520 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700	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
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	57	100	97
PCGF2	No OMIM phenotype	600346	46	97	93
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	79	100	97
PDE4D	{Stroke, susceptibility to, 1}, 606799 Acrodysostosis 2, with or without hormone resistance, 614613	600129	64	99	95
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	64	99	92
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
PEX1	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Heimler syndrome 1, 234580	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
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	603164	58	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	93	100	100
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617	601498	71	95	91
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879	601757	68	100	89
PGAP1	Mental retardation, autosomal recessive 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, autosomal recessive, 615414	602978	73	97	93
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	31	98	73
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263	300560	52	99	90
PHGDH	Phosphoglycerate dehydrogenase deficiency, 601815 Neu-Laxova syndrome 1, 256520	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
PIGG	Mental retardation, autosomal recessive 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	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398	610272	73	100	98
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	102	100	100
PIK3CA	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Nonsmall cell lung cancer, somatic, 211980 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108	171834	71	100	98
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	70	90	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953	603604	88	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	76	100	99
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	605031	53	92	69
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920	300401	71	100	99
PLXND1	No OMIM phenotype	604282	55	95	89
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	93	100	100
PNKP	Microcephaly, seizures, and developmental delay, 613402 Ataxia-oculomotor apraxia 4, 616267	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	Progressive external ophthalmoplegia, autosomal recessive 1, 258450 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459	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
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	606822	82	100	95
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	71	100	97
PPFIA4	No OMIM phenotype	603145	44	98	89
PPM1D	Breast cancer, 114480	605100	88	100	100
PPOX	Porphyria variegata, 176200	600923	69	100	100
PPP1CB	No OMIM phenotype	600590	38	98	81
PPP2R1A	Mental retardation, autosomal dominant 36, 616362	605983	61	100	96
PPP2R5D	Mental retardation, autosomal dominant 35, 616355	601646	58	100	94
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	81	100	100
PQBP1	Renpenning syndrome, 309500	300463	62	100	100
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	51	95	83
PRPS1	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500	311850	59	100	100
PRSS12	Mental retardation, autosomal recessive 1, 249500	606709	82	100	98
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722	176801	98	100	100
PSMD12	No OMIM phenotype	604450	32	83	57
PTCH1	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828	601309	88	99	96
PTCHD1	{Autism, susceptibility to, X-linked 4}, 300830	300828	54	100	99
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	46	100	94
PTEN	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355	601728	57	77	73

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PTF1A	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935	607194	75	95	84
PTGR1	No OMIM phenotype	601274	31	97	74
PTPN11	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250	176876	68	99	99
PUF60	Verheij syndrome, 615583	604819	71	97	95
PURA	Mental retardation, autosomal dominant 31, 616158	600473	87	98	86
PUS1	Mitochondrial myopathy and sideroblastic anemia 1, 600462	608109	71	100	98
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, 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
RAB18	Warburg micro syndrome 3, 614222	602207	59	97	91
RAB27A	Griscelli syndrome, type 2, 607624	603868	83	100	100
RAB39B	Mental retardation, X-linked 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	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554 Cardiomyopathy, dilated, 1NN, 615916	164760	82	100	98
RAI1	Smith-Magenis syndrome, 182290	607642	91	100	100
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	71	100	95
RBBP8	Pancreatic carcinoma, somatic Seckel syndrome 2, 606744 Jawad syndrome, 251255	604124	64	100	94
RBM10	TARP syndrome, 311900	300080	42	98	88
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	87	100	100
RELN	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436	600514	91	100	98
REV3L	No OMIM phenotype	602776	55	97	88
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	62	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RHEB	No OMIM phenotype	601293	14	59	24
RIT1	Noonan syndrome 8, 615355	609591	54	100	98
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	82	100	100
RMRP	Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460 Anauxetic dysplasia, 607095	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
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	57	100	97
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	63	100	98
RPGRIP1L	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360	610937	77	97	95
RPL10	{Autism, susceptibility to, X-linked 5}, 300847	312173	68	100	100
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844	300075	40	96	81
RTEL1	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	58	97	87
RTTN	Polymicrogyria with seizures, 614833	610436	71	99	96
RUBCN	?Spinocerebellar ataxia, autosomal recessive 15, 615705	613516	No coverage data		
SALL1	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 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
SC5D	Lathosterolosis, 607330	602286	86	100	100
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403	182389	100	100	99
SCN2A	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721	182390	100	100	99
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558	600702	97	100	99
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	94	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SDHA	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165	600857	43	77	64
SEMA3E	?CHARGE syndrome, 214800	608166	72	100	100
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	69	100	99
SETBP1	Schinzel-Giedion midface retraction syndrome, 269150 Mental retardation, autosomal dominant 29, 616078	611060	74	98	97
SETD1A	No OMIM phenotype	611052	57	97	91
SETD2	Luscan-Lumish syndrome, 616831	612778	75	100	98
SETD5	Mental retardation, autosomal dominant 23, 615761	615743	57	99	96
SF1	No OMIM phenotype	601516	36	83	69
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900	605270	86	94	94
SHANK2	{Autism susceptibility 17}, 613436	603290	81	100	100
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	56	93	82
SHH	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160	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
SIL1	Marinesco-Sjogren syndrome, 248800	608005	85	100	100
SIN3A	No OMIM phenotype	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
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	{?Schizophrenia susceptibility 18}, 615232 Dicarboxylic aminoaciduria, 222730	133550	95	100	100
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	600229	62	99	90
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	63	100	100
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 Dystonia 9, 601042 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	104	100	100
SLC33A1	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482	603690	62	100	96
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	29	98	80
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	30	94	67
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	103	100	100
SLC39A12	No OMIM phenotype	608734	39	97	81
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	71	100	99
SLC6A17	Mental retardation, autosomal recessive 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, X-linked syndromic, Christianson type, 300243	300231	50	100	96
SMAD4	Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900	600993	76	100	98
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	78	98	96
SMARCA4	{Rhabdoid tumor predisposition syndrome 2}, 613325 Mental retardation, autosomal dominant 16, 614609	603254	91	100	100
SMARCB1	Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 Mental retardation, autosomal dominant 15, 614608 {Schwannomatosis-1, susceptibility to}, 162091	601607	123	100	100
SMARCC2	No OMIM phenotype	601734	42	96	83
SMARCE1	{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, X-linked, Snyder-Robinson type, 309583	300105	39	95	94
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	99	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	88	100	100
SNRNP70	No OMIM phenotype	180740	49	99	94
SNX14	Spinocerebellar ataxia, autosomal recessive 20, 616354	616105	27	80	52
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	79	98	94
SON	No OMIM phenotype	182465	55	91	80
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	65	100	95
SOX10	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136	602229	45	99	87
SOX11	Mental retardation, autosomal dominant, 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, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000	313430	34	99	76
SOX5	Lamb-Shaffer syndrome, 616803	604975	72	100	100
SPG11	Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668	610844	73	100	99
SPRED1	Legius syndrome, 611431	609291	67	100	95
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	87	100	100
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
ST3GAL3	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006	606494	111	100	100
ST3GAL5	Amish infantile epilepsy 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, autosomal recessive, 612703	181590	81	100	97
STRA6	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 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
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	38	86	75
SUOX	Sulfite oxidase deficiency, 272300	606887	104	100	100
SURF1	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684	185620	71	88	88
SUZ12	No OMIM phenotype	606245	54	98	83
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491	313440	37	86	67
SYNCRIP	No OMIM phenotype	616686	30	79	54
SYNE1	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998	608441	87	100	100
SYNGAP1	Mental retardation, autosomal dominant 5, 612621	603384	55	95	82
SYP	Mental retardation, X-linked 96, 300802	313475	48	100	100
SYT1	No OMIM phenotype	185605	52	96	88
SYT14	Spinocerebellar ataxia, autosomal recessive 11, 614229	610949	92	93	89
TAF2	Mental retardation, autosomal recessive 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	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338 DOOR syndrome, 220500 Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044	613577	92	100	100
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000	612655	39	95	85
TBCE	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410	604934	80	99	91
TBR1	No OMIM phenotype	604616	62	100	95
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Velocardiofacial syndrome, 192430 Tetralogy of Fallot, 187500	602054	55	79	66
TCF12	Craniosynostosis 3, 615314	600480	51	100	94
TCF20	No OMIM phenotype	603107	60	100	99
TCF4	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267	602272	73	100	98
TCF7L2	{Diabetes mellitus, type 2, susceptibility to}, 125853	602228	84	100	99
TECR	Mental retardation, autosomal recessive 14, 614020	610057	68	99	94
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	100	100	100
TGIF1	Holoprosencephaly-4, 142946	602630	97	100	100
TH	Segawa syndrome, recessive, 605407	191290	55	100	95
THOC6	Beaulieu-Boycott-Innes syndrome, 613680	615403	101	100	100
THRB	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650	190160	89	100	100
TIMM8A	Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150	300356	42	100	100
TLK2	No OMIM phenotype	608439	31	88	67
TM4SF20	{Specific language impairment 5}, 615432	615404	61	100	100
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614123	60	100	98
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	76	100	100
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	56	98	93
TMEM237	Joubert syndrome 14, 614424	614423	80	100	99
TMEM5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	106	100	99
TMEM67	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Nephronophthisis 11, 613550	609884	51	99	90
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency, 300872	300777	42	98	87
TMPRSS7	No OMIM phenotype	NA	47	100	92
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 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, autosomal recessive 13, 613192	611966	85	100	99
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700	606609	129	100	100
TRIM32	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988	602290	77	100	100
TRIO	No OMIM phenotype	601893	86	99	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TRIP12	No OMIM phenotype	604506	75	100	99
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	616013	46	99	83
TSC1	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341	605284	94	100	99
TSC2	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690	191092	94	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	96	96
TSPAN7	Mental retardation, X-linked 58, 300210	300096	40	100	85
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	64	100	90
TTI2	Mental retardation, autosomal recessive 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
TUBB4A	Dystonia 4, torsion, autosomal dominant, 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, autosomal recessive, 3, 616335	609610	42	96	82
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270	610053	94	100	100
TUSC3	Mental retardation, autosomal recessive 7, 611093	601385	74	100	100
TWIST1	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750	601622	79	100	86
UBE2A	Mental retardation, X-linked 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
UPB1	Beta-ureidopropionase deficiency, 613161	606673	108	100	100
UPF3B	Mental retardation, X-linked, syndromic 14, 300676	300298	32	99	83
UQCC2	?Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	51	100	91
USP18	No OMIM phenotype	607057	58	96	94
USP7	No OMIM phenotype	602519	33	85	63

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
USP9X	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 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
VPS13B	Cohen syndrome, 216550	607817	83	100	99
VPS13C	No OMIM phenotype	608879	37	90	72
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	78	100	96
WAC	Desanto-Shinawi syndrome, 616708	615049	48	97	84
WDR13	No OMIM phenotype	300512	56	100	98
WDR19	Nephronophthisis 13, 614377 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Senior-Loken syndrome 8, 616307	608151	73	100	98
WDR4	No OMIM phenotype	605924	79	100	99
WDR45	Neurodegeneration with brain iron accululation 5, 300894	300526	32	89	78
WDR62	Microcephaly 2, primary, autosomal recessive, 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	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 ?Cataract 41, 116400	606201	102	100	100
WWOX	Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211	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	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777	608124	75	94	88
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 Coloboma, ocular, 120433	606608	40	86	73
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, autosomal dominant 22, 612337	608433	103	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ZBTB20	Primrose syndrome, 259050	606025	91	100	99
ZDHHC15	?Mental retardation, X-linked 91, 300577	300576	38	100	94
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799	300646	44	100	90
ZEB2	Mowat-Wilson syndrome, 235730	605802	81	100	100
ZFYVE26	Spastic paraplegia 15, autosomal recessive, 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, autosomal dominant 30, 616083	608668	43	98	88
ZNF292	No OMIM phenotype	616213	54	97	89
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, X-linked 97, 300803	314990	39	100	92
ZNF81	Mental retardation, X-linked 45, 300498	314998	36	100	92

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "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