Whole Exome Sequencing
Gene package Intellectual disability, version 8.1, 25-7-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
A2ML1	No OMIM phenotype	610627	57	100	99	91
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	78	100	99	93
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	56	100	96	86
ABAT	GABA-transaminase deficiency, 613163	137150	95	100	99	94
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	55	100	98	90
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	75	85	76	72
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	98	100	100	98
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	62	100	100	95
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	163	100	100	100
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	88	100	100	99
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	138	100	97	94
ACOT9	No OMIM phenotype	300862	30	100	85	47
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	95	100	100	98
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	144	100	100	99

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ACSL4	Mental retardation, X-linked 63, 300387	300157	49	100	98	83
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	208	100	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717	102560	174	100	100	100
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	65	100	100	96
ACY1	Aminoacylase 1 deficiency, 609924	104620	99	100	100	100
ADA2	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	91	100	100	97
ADAM22	?Epileptic encephalopathy, early infantile, 61, 617933	603709	58	100	98	86
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	79	100	100	98
ADAT3	Mental retardation, autosomal recessive 36, 615286	615302	126	100	100	100
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	112	100	100	99
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	44	100	91	70
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	66	100	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	78	100	99	92
AFF2	Mental retardation, X-linked, FRAXE type, 309548	300806	46	100	98	82
AFF4	CHOPS syndrome, 616368	604417	59	100	98	89
AFG3L2	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246	604581	65	100	95	87
AGA	Aspartylglucosaminuria, 208400	613228	62	100	99	92
AGAP2	No OMIM phenotype	605476	115	100	99	96
AGO1	No OMIM phenotype	606228	71	100	100	98
AGO2	No OMIM phenotype	606229	77	99	95	87
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	128	100	100	99
AGTR2	No OMIM phenotype	300034	48	100	100	98
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	143	96	96	96
AHDC1	Xia-Gibbs syndrome, 615829	615790	133	100	99	97
AHI1	Joubert syndrome 3, 608629	608894	58	100	96	82
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614	300169	45	100	95	78
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	69	100	100	95
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	119	100	100	100

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AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500	164730	136	100	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	59	100	97	82
ALDH18A1	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586	138250	68	100	100	95
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	58	100	98	92
ALDH4A1	Hyperprolinemia, type II, 239510	606811	115	100	100	100
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	61	100	97	91
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	59	91	79	72
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	61	100	100	97
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	152	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	41	100	93	68
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	70	100	100	97
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	78	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	63	100	100	90
ALG8	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874	608103	54	100	99	88
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	54	100	99	89
ALMS1	Alstrom syndrome, 203800	606844	84	100	99	97
ALX1	?Frontonasal dysplasia 3, 613456	601527	68	100	100	98
ALX4	{Craniosynostosis 5, susceptibility to}, 615529 Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597	605420	137	100	100	100
AMMECR1	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990	300195	46	99	85	73
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686	102771	136	100	100	100
AMT	Glycine encephalopathy, 605899	238310	108	100	100	100
ANK3	?Mental retardation, autosomal recessive, 37, 615493	600465	60	100	100	96
ANKEF1	No OMIM phenotype	No id	67	100	99	94

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ANKH	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	605145	79	100	100	99
ANKLE2	?Microcephaly 16, primary, autosomal recessive, 616681	616062	96	100	98	94
ANKRD11	KBG syndrome, 148050	611192	111	100	100	98
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728	613726	49	100	96	81
ANTXR1	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089	606410	49	98	96	84
AP1S1	MEDNIK syndrome, 609313	603531	73	100	97	96
AP1S2	Mental retardation, X-linked syndromic 5, 304340	300629	40	100	87	62
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	57	100	96	80
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276	602166	97	100	100	98
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066	607245	70	100	100	98
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450	607244	55	100	99	90
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936	602296	117	100	100	99
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067	607243	38	100	97	74
APC2	?Sotos syndrome 3, 617169	612034	116	100	97	94
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	67	100	98	87
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	74	100	99	92
ARG1	Argininemia, 207800	608313	63	100	100	92
ARHGAP31	Adams-Oliver syndrome 1, 100300	610911	89	100	100	98
ARHGAP4	No OMIM phenotype	300023	70	100	99	96
ARHGEF33	No OMIM phenotype	No id	66	99	94	85
ARHGEF6	?Mental retardation, X-linked 46, 300436	300267	39	100	93	75
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	44	100	96	71
ARID1A	Coffin-Siris syndrome 2, 614607	603024	99	100	99	95
ARID1B	Coffin-Siris syndrome 1, 135900	614556	87	100	99	97
ARID2	Coffin-Siris syndrome 6, 617808	609539	65	100	99	92
ARL13B	Joubert syndrome 8, 612291	608922	53	100	99	90
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	35	96	93	71
ARNT2	?Webb-Dattani syndrome, 615926	606036	81	100	100	94
ARSA	Metachromatic leukodystrophy, 250100	607574	136	100	100	100
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950	300180	69	100	97	83

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ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	46	91	81	72
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	57	100	98	85
ASCL1	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880	100790	230	100	100	100
ASL	Argininosuccinic aciduria, 207900	608310	110	100	100	98
ASNS	Asparagine synthetase deficiency, 615574	108370	64	100	99	90
ASPA	Canavan disease, 271900	608034	49	100	98	86
ASPM	Microcephaly 5, primary, autosomal recessive, 608716	605481	61	100	100	95
ASS1	Citrullinemia, 215700	603470	111	100	97	88
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	94	100	100	98
ASXL2	Shashi-Pena syndrome, 617190	612991	79	100	98	93
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	59	99	98	95
ATAD3A	Harel-Yoon syndrome, 617183	612316	128	99	98	97
ATAD3B	No OMIM phenotype	612317	130	99	98	95
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	104	100	98	96
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	54	100	99	93
ATN1	Dentatorubro-pallidoluysian atrophy, 125370	607462	131	100	100	98
ATP1A2	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	119	100	100	100
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	134	100	100	100
ATP2A2	Acrokeratosis verruciformis, 101900 Darier disease, 124200	108740	83	100	100	99
ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911	300556	41	100	87	60
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	70	100	99	94
ATP6V1B2	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455	606939	68	100	100	98

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ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489	300011	40	100	95	71
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	62	100	98	89
ATPAF2	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	68	100	100	98
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600	601215	79	100	97	85
ATRIP	No OMIM phenotype	606605	97	100	100	96
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580	300032	33	100	90	59
AUH	3-methylglutaconic aciduria, type I, 250950	600529	84	100	100	95
AUTS2	Mental retardation, autosomal dominant 26, 615834	607270	95	100	99	95
AVPR2	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	300538	77	100	100	98
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	52	100	99	89
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	66	79	75	72
B3GLCT	Peters-plus syndrome, 261540	610308	60	100	95	79
B4GALNT1	Spastic paraplegia 26, autosomal recessive, 609195	601873	108	100	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	98	100	100	98
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	604327	129	100	100	98
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	160	100	100	100
BBS1	Bardet-Biedl syndrome 1, 209900	209901	109	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	57	100	100	99
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	98
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	68	100	100	93
BBS4	Bardet-Biedl syndrome 4, 615982	600374	68	100	99	91
BBS5	Bardet-Biedl syndrome 5, 615983	603650	57	100	97	83
BBS7	Bardet-Biedl syndrome 7, 615984	607590	56	100	99	92
BBS9	Bardet-Biedl syndrome 9, 615986	607968	49	96	93	85
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	77	100	100	95
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	146	100	100	99
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	56	100	98	89
BCL11A	Dias-Logan syndrome, 617101	606557	106	100	100	99
BCOR	Microphthalmia, syndromic 2, 300166	300485	69	100	98	91

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BCORL1	No OMIM phenotype	300688	100	100	100	96
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	165	100	100	100
BLM	Bloom syndrome, 210900	604610	71	100	99	92
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	100	99	90
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	121	100	100	100
BRF1	Cerebellofaciodental syndrome, 616202	604902	106	100	100	99
BRPF1	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333	602410	119	100	100	99
BRWD3	Mental retardation, X-linked 93, 300659	300553	39	100	94	69
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	90	100	100	99
BTD	Biotinidase deficiency, 253260	609019	82	100	100	98
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	60	100	98	89
c12orf4	No OMIM phenotype	616082	67	100	99	91
C12orf57	Temtamy syndrome, 218340	615140	140	100	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035	613541	69	100	100	99
C1orf167	No OMIM phenotype	No id	97	100	98	95
C2CD3	?Orofaciodigital syndrome XIV, 615948	615944	67	100	100	96
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730	611492	80	100	100	99
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	164	100	100	100
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	66	100	99	86

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CACNA1A	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086	601011	80	100	98	92
CACNA1C	Brugada syndrome 3, 611875 Timothy syndrome, 601005	114205	108	100	99	98
CACNA2D1	No OMIM phenotype	114204	53	100	98	86
CACNG2	?Mental retardation, autosomal dominant 10, 614256	602911	92	100	100	98
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	109	100	100	100
CAMK2A	Mental retardation, autosomal dominant 53, 617798	114078	69	100	98	93
CAMK2B	Mental retardation, autosomal dominant 54, 617799	607707	88	100	98	96
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	142	100	99	96
CAPN10	{Diabetes mellitus, noninsulin-dependent 1}, 601283	605286	107	100	100	100
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	44	100	94	67
CBL	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	69	100	100	99
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	122	100	100	100
CC2D1A	Mental retardation, autosomal recessive 3, 608443	610055	124	100	100	100
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	56	100	99	90
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	71	100	98	95
CCDC13	No OMIM phenotype	No id	77	100	100	98
CCDC14	No OMIM phenotype	617147	66	100	99	91
CCDC174	Hypotonia, infantile, with psychomotor retardation, 616816	616735	54	100	95	77
CCDC22	Ritscher-Schinzel syndrome 2, 300963	300859	73	100	94	92
CCDC78	?Centronuclear myopathy 4, 614807	614666	121	100	100	100
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053	611204	104	100	99	97
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	123833	98	100	100	97
CDC5L	No OMIM phenotype	602868	47	100	97	85
CDH15	Mental retardation, autosomal dominant 3, 612580	114019	114	100	100	99
CDK16	No OMIM phenotype	311550	61	100	100	95
CDK5	?Lissencephaly 7 with cerebellar hypoplasia, 616342	123831	102	100	100	100

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CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804	608201	63	100	98	88
CDK6	?Microcephaly 12, primary, autosomal recessive, 616080	603368	70	100	100	94
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	48	100	95	79
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	80	92	85	78
CDON	Holoprosencephaly 11, 614226	608707	76	100	100	96
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676	609279	64	100	100	95
CEP104	Joubert syndrome 25, 616781	616690	64	100	97	88
CEP135	Microcephaly 8, primary, autosomal recessive, 614673	611423	72	100	97	86
CEP152	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823	613529	57	100	97	89
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	59	100	96	81
CEP41	Joubert syndrome 15, 614464	610523	61	100	99	92
CEP63	?Seckel syndrome 6, 614728	614724	61	100	97	84
CEP89	No OMIM phenotype	615470	60	100	96	84
CHAMP1	Mental retardation, autosomal dominant 40, 616579	616327	117	100	100	100
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	60	100	99	91
CHD3	No OMIM phenotype	602120	75	98	95	89
CHD4	Sifrim-Hitz-Weiss syndrome, 617159	603277	60	100	100	96
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	74	100	99	94
CHD8	{Autism, susceptibility to, 18}, 615032	610528	76	100	100	95
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	104	100	100	100
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	91	100	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	132	100	100	96
CIC	Mental retardation, autosomal dominant 45, 617600	612082	119	100	100	98
CIT	Microcephaly 17, primary, autosomal recessive, 617090	605629	85	100	99	94
CKAP2L	Filippi syndrome, 272440	616174	69	100	100	99
CLCN4	Mental retardation, X-linked 49/15, 300114	302910	64	100	100	92
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	147	100	100	100
CLIC2	?Mental retardation, X-linked, syndromic 32, 300886	300138	32	100	85	56

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CLIP1	No OMIM phenotype	179838	72	100	98	90
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	101	100	100	99
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	62	100	100	99
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	117	100	100	99
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	123	100	100	100
CLP1	Pontocerebellar hypoplasia, type 10, 615803	608757	83	100	100	100
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	110	100	100	99
CLTC	Mental retardation, autosomal dominant 56, 617854	118955	55	100	97	90
CNKSR2	Mental retardation, X-linked, syndromic, Houge type, 301008	300724	46	100	93	79
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	148	100	100	97
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	60	100	100	96
COASY	Neurodegeneration with brain iron accumulation 6, 615643	609855	129	100	100	100
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	89	100	99	95
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	75	100	100	96
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	58	100	98	87
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	58	100	98	82
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	77	100	100	96
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	120	100	100	100
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	53	100	98	84
COL18A1	Knobloch syndrome, type 1, 267750	120328	138	100	100	98
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 Schizencephaly, 269160	120130	80	100	100	96
COL4A2	{Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 2, 614483	120090	97	100	100	98
COL4A3BP	Mental retardation, autosomal dominant 34, 616351	604677	55	99	94	82

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COLEC11	3MC syndrome 2, 265050	612502	158	100	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	99	89
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276	612898	112	100	100	100
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	125	100	100	99
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	109	100	100	100
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	131	100	100	96
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	56	100	97	87
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	87	100	100	100
CPLANE1	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	59	100	98	90
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	52	100	99	91
CRADD	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499	603454	139	100	100	100
CRB2	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	609720	120	100	100	100
CRBN	Mental retardation, autosomal recessive 2, 607417	609262	60	100	98	84
CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	82	100	98	93
CRLF1	Cold-induced sweating syndrome 1, 272430	604237	104	95	90	87
CSNK2A1	Okur-Chung neurodevelopmental syndrome, 617062	115440	63	100	92	80
CSPP1	Joubert syndrome 21, 615636	611654	73	100	99	93
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	75	100	100	100
CTBP1	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915	602618	90	96	87	82
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	90	100	100	99
CTCF	Mental retardation, autosomal dominant 21, 615502	604167	88	100	100	100
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	119	99	91	88
CTNNB1	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	52	100	99	94
CTNND1	Blepharocheilodontic syndrome 2, 617681	601045	61	100	99	93
CTNND2	No OMIM phenotype	604275	81	96	92	85

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CTSA	Galactosialidosis, 256540	613111	118	100	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	138	100	100	100
CTTNBP2	No OMIM phenotype	609772	86	100	99	94
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	71	100	99	90
CUL3	Pseudohypoaldosteronism, type IIE, 614496	603136	62	100	94	83
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354	300304	44	100	93	74
CWF19L1	Spinocerebellar ataxia, autosomal recessive 17, 616127	616120	51	100	98	83
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	120	100	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	126	100	100	100
CYP2U1	Spastic paraplegia 56, autosomal recessive, 615030	610670	58	100	100	95
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	148	100	100	100
DAB1	Spinocerebellar ataxia 37, 615945	603448	61	100	100	95
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	128239	136	100	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	53	100	100	92
DBT	Maple syrup urine disease, type II, 248600	248610	99	100	100	98
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	66	100	98	91
DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600	120470	58	100	100	95
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	118	100	100	100
DCPS	Al-Raqad syndrome, 616459	610534	112	100	100	100
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067	300121	48	100	97	87
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	75	100	97	89
DDHD2	Spastic paraplegia 54, autosomal recessive, 615033	615003	66	100	100	94
DDX11	Warsaw breakage syndrome, 613398	601150	250	100	100	100
DDX3X	Mental retardation, X-linked 102, 300958	300160	67	100	99	96
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828	602635	86	100	96	87
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281	617278	54	100	97	89
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	76	100	99	94
DHCR24	Desmosterolosis, 602398	606418	113	100	100	100
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	98	100	100	100
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	74	100	100	97

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DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	76	100	100	96
DIAPH1	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	77	100	98	88
DIP2B	Mental retardation, FRA12A type, 136630	611379	57	100	99	93
DKC1	Dyskeratosis congenita, X-linked, 305000	300126	43	100	97	79
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	66	100	100	94
DLG3	Mental retardation, X-linked 90, 300850	300189	57	100	96	80
DLG4	No OMIM phenotype	602887	110	100	100	100
DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	37	100	92	67
DMPK	Myotonic dystrophy 1, 160900	605377	109	100	100	99
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	60	100	94	85
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	65	100	100	89
DNM1	Epileptic encephalopathy, early infantile, 31, 616346	602377	122	100	100	95
DNMT3A	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879	602769	111	100	100	99
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	105	100	100	97
DOCK6	Adams-Oliver syndrome 2, 614219	614194	101	100	98	95
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859	615730	49	100	98	85
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700	611432	64	100	98	91
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	123	100	100	100
DONSON	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230	611428	39	100	91	71
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	66	100	100	97
DPF2	Coffin-Siris syndrome 7, 618027	601671	72	100	98	93
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901	603527	115	100	100	100
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	70	93	89	83
DPP6	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956	126141	72	100	96	85
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	54	100	99	90
DPYS	Dihydropyrimidinuria, 222748	613326	63	100	100	96
DST	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653	113810	57	100	98	91

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DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	56	100	96	81
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	100	96
DYRK1A	Mental retardation, autosomal dominant 7, 614104	600855	64	100	100	94
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960	300205	76	100	100	100
EDC3	?Mental retardation, autosomal recessive 50, 616460	609842	103	100	100	100
EDNRB	ABCD syndrome, 600501 {Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580	131244	86	100	100	99
EDRF1	No OMIM phenotype	No id	56	100	99	90
EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393	602959	161	100	100	100
EFCAB1	No OMIM phenotype	No id	55	100	100	89
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536	603892	76	100	100	95
EHMT1	Kleefstra syndrome 1, 610253	607001	119	99	99	99
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	62	100	99	91
EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305	608546	61	100	100	92
EIF4G1	{Parkinson disease 18}, 614251	600495	89	100	100	99
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731	605367	76	100	100	96
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	61	100	100	93
ELP2	Mental retardation, autosomal recessive 58, 617270	616054	66	100	100	95
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	110	100	100	97
EMG1	Bowen-Conradi syndrome, 211180	611531	63	100	100	100
EML1	Band heterotopia, 600348	602033	64	100	99	91
EMX2	Schizencephaly, 269160	600035	143	100	100	100
ENTPD1	Spastic paraplegia 64, autosomal recessive, 615683	601752	57	100	99	88
EOMES	No OMIM phenotype	604615	84	100	100	100
EP300	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684	602700	89	100	99	95
EPB41L1	?Mental retardation, autosomal dominant 11, 614257	602879	114	100	100	99
EPG5	Vici syndrome, 242840	615068	61	100	99	93
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	69	100	100	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ERCC2	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	91	100	99	97
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	133510	70	100	100	96
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	133530	75	100	100	96
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	74	100	99	94
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	74	100	97	86
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225	611605	59	100	99	92
ERMARD	?Periventricular nodular heterotopia 6, 615544	615532	59	100	99	90
ESCO2	Roberts syndrome, 268300 SC phocomelia syndrome, 269000	609353	51	100	99	92
ETFB	Glutaric acidemia IIB, 231680	130410	102	100	100	100
ETHE1	Ethylmalonic encephalopathy, 602473	608451	108	100	100	93
EXOSC2	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	602238	61	100	100	95
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	93	100	100	88
EZH2	Weaver syndrome, 277590	601573	69	100	100	94
FA2H	Spastic paraplegia 35, autosomal recessive, 612319	611026	73	100	100	95
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	54	100	100	95
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154	616107	63	100	100	92
FAT2	Spinocerebellar ataxia 45, 617769	604269	85	100	100	98
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	78	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	175	100	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	66	100	99	94
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	605654	59	100	100	96
FBXO31	?Mental retardation, autosomal recessive 45, 615979	609102	98	100	99	96
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	65	100	99	95
FGF12	Epileptic encephalopathy, early infantile, 47, 617166	601513	48	100	95	76
FGF14	Spinocerebellar ataxia 27, 609307	601515	58	100	100	96
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	94	100	100	98
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579	176943	63	100	99	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	112	100	100	99
FGL1	No OMIM phenotype	605776	48	100	98	85
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	70	99	94	86
FIBP	Thauvin-Robinet-Faivre syndrome, 617107	608296	92	100	100	99
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	53	100	99	88
FIGN	No OMIM phenotype	605295	77	100	100	99
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	130	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	72	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	98	100	100	99
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	87	100	99	93
FMN2	Mental retardation, autosomal recessive 47, 616193	606373	86	99	92	86
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	34	100	89	59
FOXG1	Rett syndrome, congenital variant, 613454	164874	103	98	91	83
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	72	100	100	96
FOXP2	Speech-language disorder-1, 602081	605317	58	100	100	93
FRAS1	Fraser syndrome 1, 219000	607830	70	100	99	94
FREM2	Fraser syndrome 2, 617666	608945	93	100	100	98
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	616305	75	100	96	85
FRMPD4	Mental retardation, X-linked 104, 300983	300838	61	100	98	87
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981	604574	39	84	71	58
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	93	99	96	93
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	73	100	100	95
FTSJ1	Mental retardation, X-linked 9/44, 309549	300499	77	100	99	90
FUCA1	Fucosidosis, 230000	612280	79	100	100	94
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	63	100	100	96
GABRB1	Epileptic encephalopathy, early infantile, 45, 617153	137190	69	100	99	93
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	85	100	99	97
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	78	100	100	96
GALE	Galactose epimerase deficiency, 230350	606953	109	100	100	100
GALT	Galactosemia, 230400	606999	146	100	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	80	100	99	95

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GAS6	No OMIM phenotype	600441	88	100	96	93
GATAD2B	Mental retardation, autosomal dominant 18, 615074	614998	61	100	100	97
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	56	100	100	93
GCDH	Glutaricaciduria, type I, 231670	608801	104	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	49	100	100	91
GCSH	?Glycine encephalopathy, 605899	238330	88	100	91	63
GDI1	Mental retardation, X-linked 41, 300849	300104	86	100	100	99
GFAP	Alexander disease, 203450	137780	79	100	100	99
GFM2	No OMIM phenotype	606544	54	100	97	86
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	82	100	100	97
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800	304040	76	100	100	98
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206	608803	92	98	88	81
GK	Glycerol kinase deficiency, 307030	300474	40	99	86	60
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	96	100	100	96
GLDC	Glycine encephalopathy, 605899	238300	61	100	97	87
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	144	100	100	98
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	102	100	100	98
GLYCTK	D-glyceric aciduria, 220120	610516	135	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	85	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	99	100	100	99

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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	168	100	100	100
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493	139311	89	100	100	96
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	600998	85	100	100	100
GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463	139320	164	100	99	97
GNB1	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973	139380	81	100	100	94
GNB5	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182	604447	60	100	99	89
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	66	100	99	91
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	54	100	98	89
GNS	Mucopolysaccharidosis type IIID, 252940	607664	60	100	100	96
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	46	100	97	79
GPHN	Molybdenum cofactor deficiency C, 615501	603930	59	100	100	93
GPT2	Mental retardation, autosomal recessive 49, 616281	138210	84	100	99	89
GRIA3	Mental retardation, X-linked 94, 300699	305915	40	100	94	75
GRID2	Spinocerebellar ataxia, autosomal recessive 18, 616204	602368	66	100	100	96
GRIK2	Mental retardation, autosomal recessive, 6, 611092	138244	79	100	100	96
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820	138249	126	100	100	100
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	89	100	100	99
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970	138252	113	100	100	98
GRIN3B	No OMIM phenotype	606651	128	92	89	83

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GRIP1	Fraser syndrome 3, 617667	604597	75	100	100	94
GRM1	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831	604473	114	100	100	98
GSE1	No OMIM phenotype	616886	97	100	100	100
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	82	100	100	98
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	44	100	100	90
GTPBP3	Combined oxidative phosphorylation deficiency 23, 616198	608536	141	100	100	100
GUCY2F	No OMIM phenotype	300041	36	100	94	69
GUSB	Mucopolysaccharidosis VII, 253220	611499	100	100	100	97
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	69	100	99	90
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738	605998	90	100	100	98
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	36	100	95	75
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	71	100	98	89
HCN1	Epileptic encephalopathy, early infantile, 24, 615871	602780	70	100	99	93
HDAC4	No OMIM phenotype	605314	104	100	100	99
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	300272	71	100	99	91
HDAC8	Cornelia de Lange syndrome 5, 300882	300269	40	100	97	80
HECTD1	No OMIM phenotype	No id	48	100	94	83
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	617245	62	100	95	85
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	86	100	100	97
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011	605109	54	100	98	88
HERC2	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	100	100	100	97
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230	601802	60	100	100	88
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	79	100	100	96
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	106	100	100	95
HIVEP2	Mental retardation, autosomal dominant 43, 616977	143054	90	100	99	97

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HLCS	Holocarboxylase synthetase deficiency, 253270	609018	85	100	100	97
HMGCL	HMG-CoA lyase deficiency, 246450	613898	78	100	100	100
HNMT	{Asthma, susceptibility to}, 600807 Mental retardation, autosomal recessive 51, 616739	605238	64	100	100	96
HNRNPH2	Mental retardation, X-linked, syndromic, Bain type, 300986	300610	64	100	100	100
HNRNPK	Au-Kline syndrome, 616580	600712	42	95	80	60
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391	602869	71	100	99	90
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	115	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	94	100	100	96
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	34	100	93	70
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	178	100	100	100
HSD17B10	HSD10 mitochondrial disease, 300438	300256	62	100	100	100
HSPA9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854	600548	62	100	98	90
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280	118190	56	100	97	81
HUWE1	Mental retardation, X-linked syndromic, Turner type, 300706	300697	45	100	92	72
IARS	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093	600709	42	100	96	79
IBA57	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451	615316	126	100	100	98
IDS	Mucopolysaccharidosis II, 309900	300823	68	100	98	84
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	127	100	99	94
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	65	100	98	76
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	74	100	99	93
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	65	100	99	92

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IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895	605489	39	99	91	72
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	59	100	99	79
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	63	100	100	99
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	77	100	100	96
IL1RAPL1	Mental retardation, X-linked 21/34, 300143	300206	42	100	96	81
IMPA1	Mental retardation, autosomal recessive 59, 617323	602064	48	99	88	66
INPP5B	No OMIM phenotype	147264	81	100	100	95
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	115	100	100	98
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404	607875	64	100	100	96
INTS1	No OMIM phenotype	611345	102	100	99	96
INTS8	No OMIM phenotype	611351	44	100	94	74
IQSEC2	Mental retardation, X-linked 1/78, 309530	300522	57	98	92	84
ISG15	Immunodeficiency 38, 616126	147571	135	100	100	100
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	77	100	99	92
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	600536	108	99	97	96
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	79	100	100	96
ITPR2	?Anhidrosis, isolated, with normal sweat glands, 106190	600144	48	100	93	77
ITSN1	No OMIM phenotype	602442	59	100	96	85
IVD	Isovaleric acidemia, 243500	607036	105	100	100	98
JAG1	Alagille syndrome 1, 118450 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Tetralogy of Fallot, 187500	601920	85	100	99	95
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	61	100	100	93
JMJD1C	No OMIM phenotype	604503	58	100	98	90
KALRN	{Coronary heart disease, susceptibility to, 5}, 608901	604605	79	100	98	93
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900	607704	89	100	100	97
KANSL1	Koolen-De Vries syndrome, 610443	612452	79	100	100	96
KAT6A	Mental retardation, autosomal dominant 32, 616268	601408	81	100	100	97

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KAT6B	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736	605880	89	100	99	95
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	133	100	100	100
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366	176262	99	100	100	100
KCNA4	No OMIM phenotype	176266	77	100	100	100
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056	600397	114	100	100	100
KCNC3	Spinocerebellar ataxia 13, 605259	176264	83	94	79	63
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500	603305	109	100	100	97
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	155	100	100	100
KCNJ11	Diabetes mellitus, transient neonatal, 3, 610582 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329	600937	124	100	100	100
KCNJ6	Keppen-Lubinsky syndrome, 614098	600877	79	100	100	99
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292	605874	121	100	100	100
KCNMA1	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446	600150	80	100	99	93
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data			
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200	602235	129	100	100	100
KCNQ3	Seizures, benign neonatal, 2, 121201	602232	98	100	100	95
KCNQ5	Mental retardation, autosomal dominant 46, 617601	607357	70	100	99	94
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959	608167	116	100	99	99
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	156	100	100	100
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features, 616728	609132	54	100	99	87
KDM4B	No OMIM phenotype	609765	113	100	100	98
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534	314690	75	100	99	95
KDM6A	Kabuki syndrome 2, 300867	300128	43	100	95	72
KDM6B	No OMIM phenotype	611577	119	100	97	94
KDSR	Erythrokeratodermia variabilis et progressiva 4, 617526	136440	58	100	100	91
KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	610178	62	100	97	91
KIAA1109	Alkuraya-Kucinskas syndrome, 617822	611565	66	100	99	93

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KIAA1586	No OMIM phenotype	No id	53	100	99	93
KIDINS220	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296	615759	51	100	96	84
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	63	100	98	86
KIF1A	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357	601255	97	100	100	98
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	69	100	100	96
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	72	100	97	85
KIF4A	?Mental retardation, X-linked 100, 300923	300521	41	100	92	65
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282	604593	59	100	98	87
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	104	99	96	93
KIRREL3	No OMIM phenotype	607761	113	100	100	100
KLHL15	Mental retardation, X-linked 103, 300982	300980	40	100	96	81
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	60	100	100	95
KMT2B	Dystonia 28, childhood-onset, 617284	606834	130	97	95	93
KMT2C	Kleefstra syndrome 2, 617768	606833	72	100	99	94
KMT2D	Kabuki syndrome 1, 147920	602113	106	100	100	99
KNL1	Microcephaly 4, primary, autosomal recessive, 604321	609173	51	100	98	93
KPTN	Mental retardation, autosomal recessive 41, 615637	615620	133	100	100	98
KRAS	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	190070	73	100	97	72
KRBOX4	No OMIM phenotype	300585	58	100	100	89

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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	85	100	100	99
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	61	100	100	93
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	76	100	99	94
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855	156225	60	100	99	92
LAMC1	No OMIM phenotype	150290	72	100	99	93
LAMC3	Cortical malformations, occipital, 614115	604349	121	100	100	99
LAMP2	Danon disease, 300257	309060	40	100	91	68
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	86	100	100	98
LARP7	Alazami syndrome, 615071	612026	56	100	97	85
LAS1L	Wilson-Turner syndrome, 309585	300964	63	100	100	91
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462	607031	67	100	99	93
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	61	100	100	100
LINS1	Mental retardation, autosomal recessive 27, 614340	610350	49	100	99	90
LMAN2L	?Mental retardation, autosomal recessive, 52, 616887	609552	63	100	97	92
LONP1	CODAS syndrome, 600373	605490	129	100	100	100
LRP2	Donnai-Barrow syndrome, 222448	600073	58	100	99	91
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	51	100	98	84
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	66	100	100	89
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	604357	162	100	100	100
MACF1	No OMIM phenotype	608271	57	100	97	89
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	84	85	80	77
MAGEC3	No OMIM phenotype	300469	58	100	94	86
MAGEL2	Schaaf-Yang syndrome, 615547	605283	91	100	99	95
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	44	100	97	81
MAN1B1	Mental retardation, autosomal recessive 15, 614202	604346	120	100	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	110	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	76	100	98	92
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	42	100	97	83

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MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	72	100	100	92
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	111	100	100	95
MAPRE2	Symmetric circumferential skin creases, congenital, 2, 616734	605789	74	100	96	84
MASP1	3MC syndrome 1, 257920	600521	104	100	100	97
MAST1	No OMIM phenotype	612256	129	100	100	100
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850	610550	100	100	100	100
MBD5	Mental retardation, autosomal dominant 1, 156200	611472	66	100	100	98
MBOAT7	Mental retardation, autosomal recessive 57, 617188	606048	95	100	100	100
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918	300294	39	100	96	79
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	60	100	99	89
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	56	100	99	90
MCM3AP	No OMIM phenotype	603294	72	100	98	93
MCOLN1	Mucolipidosis IV, 252650	605248	121	100	100	100
MCOLN3	No OMIM phenotype	607400	40	99	91	75
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200	607117	76	94	94	88
MDH2	Epileptic encephalopathy, early infantile, 51, 617339	154100	85	100	100	100
MECP2	{Autism susceptibility, X-linked 3}, 300496 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	106	100	100	94
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	89	100	100	98
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450	300188	57	100	99	92
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808	608771	64	100	100	94
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	81	100	100	96
MED23	Mental retardation, autosomal recessive 18, 614249	605042	54	100	99	90
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589	610197	121	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	79	100	98	92
MEIS2	Cleft palate, cardiac defects, and mental retardation, 600987	601740	81	100	100	97
METTL23	Mental retardation, autosomal recessive 44, 615942	615262	57	100	100	95
MFSD2A	Microcephaly 15, primary, autosomal recessive, 616486	614397	79	100	100	97
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	56	100	99	89
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	94	100	100	100
MICU1	Myopathy with extrapyramidal signs, 615673	605084	46	100	96	75
MID1	Opitz GBBB syndrome, type I, 300000	300552	74	100	97	85
MID2	?Mental retardation, X-linked 101, 300928	300204	46	100	93	75
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	67	100	100	98
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	112	100	100	98
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	76	100	98	90
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	69	100	98	90
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	67	100	100	97
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	115	100	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	55	100	99	81
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	108	100	100	99
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	60	100	100	94
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	117	100	100	100
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	78	100	100	98
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219	603785	60	100	99	91
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	59	100	100	98
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582	607118	56	100	99	87
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	70	100	100	91
MSL2	No OMIM phenotype	614802	67	100	100	98
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947	611766	67	100	98	83
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	95	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MTMR9	No OMIM phenotype	606260	74	100	99	94
MTOR	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638	601231	87	100	100	95
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	69	100	99	93
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	81	100	99	96
MMUT	Methylmalonic aciduria, mut(0) type, 251000	609058	62	100	99	91
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	96	100	100	98
MYCN	Feingold syndrome 1, 164280	164840	153	100	100	100
MYH9	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100	160775	121	100	100	99
MYO5A	Griscelli syndrome, type 1, 214450	160777	62	100	98	90
MYT1L	Mental retardation, autosomal dominant 39, 616521	613084	88	100	100	96
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855	300013	82	100	100	96
NAA15	Mental retardation, autosomal dominant 50, 617787	608000	70	100	98	89
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393	610672	156	100	100	100
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	114	100	100	99
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	114	100	97	93
NALCN	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	611549	60	100	99	93
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	57	100	99	93
NARS2	Combined oxidative phosphorylation deficiency 24, 616239	612803	47	99	96	84
NAT10	No OMIM phenotype	609221	73	100	98	93
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	59	100	97	81
NCOR2	No OMIM phenotype	600848	84	99	94	86
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	99	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NDP	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600	300658	73	100	100	100
NDST1	Mental retardation, autosomal recessive 46, 616116	600853	135	100	100	100
NDUFA1	Mitochondrial complex I deficiency, 252010	300078	99	100	100	100
NDUFA11	Mitochondrial complex I deficiency, 252010	612638	115	100	100	100
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000	614530	63	100	100	97
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	98	100	100	100
NDUFAF3	Mitochondrial complex I deficiency, 252010	612911	125	100	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010	612360	63	100	99	89
NDUFS1	Mitochondrial complex I deficiency, 252010	157655	61	100	99	87
NDUFS2	Mitochondrial complex I deficiency, 252010	602985	71	100	100	96
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	603846	102	100	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010	602694	69	100	100	97
NDUFS6	Mitochondrial complex I deficiency, 252010	603848	69	100	100	97
NDUFS7	Leigh syndrome, 256000	601825	114	100	100	99
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	131	100	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010	161015	122	100	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010	600532	45	100	93	75
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	72	100	100	98
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	600644	102	100	100	100
NEDD4L	Periventricular nodular heterotopia 7, 617201	606384	62	100	98	91
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	143	100	100	100
NEXMIF	Mental retardation, X-linked 98, 300912	300524	39	100	99	81
NF1	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	47	97	87	74
NFATC1	No OMIM phenotype	600489	137	100	100	99
NFE2L2	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744	600492	57	100	97	92
NFIA	Brain malformations with or without urinary tract defects, 613735	600727	72	100	100	97
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	136	100	100	99
NGLY1	Congenital disorder of deglycosylation, 615273	610661	68	100	100	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NHS	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350	300457	46	100	95	83
NID1	No OMIM phenotype	131390	104	100	100	100
NIN	?Seckel syndrome 7, 614851	608684	72	100	98	90
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	55	100	96	84
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550	600635	89	100	100	100
NLGN3	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425	300336	80	100	100	97
NLGN4X	{Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495 Mental retardation, X-linked, 300495	300427	159	100	100	100
NLRP3	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900	606416	108	100	100	100
NONO	Mental retardation, X-linked, syndromic 34, 300967	300084	41	97	85	69
NOVA2	No OMIM phenotype	601991	105	97	94	91
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	74	100	100	97
NPC2	Niemann-pick disease, type C2, 607625	601015	87	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	50	100	96	82
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118	600928	80	100	98	94
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	169	100	100	100
NRAS	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470	164790	53	100	100	95
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	88	100	99	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NSD1	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	67	100	99	94
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	62	100	98	88
NSUN2	Mental retardation, autosomal recessive 5, 611091	610916	79	100	98	91
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	115	100	100	100
NUP62	Striatonigral degeneration, infantile, 271930	605815	100	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	54	100	94	80
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	78	100	100	99
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	35	100	91	67
ODC1	{Colonic adenoma recurrence, reduced risk of}, 114500	165640	59	100	100	98
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	37	100	89	63
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	42	100	91	67
ORC1	Meier-Gorlin syndrome 1, 224690	601902	76	100	99	93
OTC	Ornithine transcarbamylase deficiency, 311250	300461	46	100	94	80
P2RX6	No OMIM phenotype	608077	106	100	97	91
PACS1	Schuurs-Hoeijmakers syndrome, 615009	607492	84	100	99	96
PACS2	No OMIM phenotype	610423	130	100	99	96
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	72	100	94	89
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	57	100	98	91
PAK3	Mental retardation, X-linked 30/47, 300558	300142	40	100	93	72
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	72	100	100	96
PANX1	No OMIM phenotype	608420	62	100	98	88
PAX1	?Otofaciocervical syndrome 2, 615560	167411	174	100	91	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	66	100	99	94
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	167415	105	100	100	95
PBX1	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641	176310	65	100	100	95
PC	Pyruvate carboxylase deficiency, 266150	608786	135	100	100	99
PCCA	Propionicacidemia, 606054	232000	58	100	99	90
PCCB	Propionicacidemia, 606054	232050	62	100	97	92
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	99	100	99	93
PCGF2	No OMIM phenotype	600346	77	100	98	95
PCLO	?Pontocerebellar hypoplasia, type 3, 608027	604918	70	100	99	96
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	121	100	100	98
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799	600129	62	100	97	91
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	41	99	93	77
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	76	100	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	51	99	94	83
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	64	100	95	81
PEPD	Prolidase deficiency, 170100	613230	92	100	100	97
PET100	Mitochondrial complex IV deficiency, 220110	614770	70	99	66	66
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	52	100	98	90
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	96	100	100	96
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	136	100	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	57	100	99	87
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	58	100	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	124	100	96	93
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	56	100	100	94
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	57	100	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	123	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	50	100	99	90
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	104	100	100	99
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	97	100	100	96
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	49	100	100	95
PGAP1	Mental retardation, autosomal recessive 42, 615802	611655	56	100	97	83
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	141	100	100	100
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	107	100	100	100
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	37	100	99	81
PHC1	?Microcephaly 11, primary, autosomal recessive, 615414	602978	153	100	100	100
PHF23	No OMIM phenotype	612910	104	100	100	100
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	42	100	95	74
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263	300560	51	100	96	73
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	135	100	100	99
PHIP	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991	612870	59	100	97	86
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	600286	111	100	99	95
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	56	100	100	93
PIGC	Glycosylphosphatidylinositol biosynthesis defect 16, 617816	601730	122	100	100	100
PIGG	Mental retardation, autosomal recessive 53, 616917	616918	92	100	100	98
PIGL	CHIME syndrome, 280000	605947	87	100	100	97
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	53	100	97	80
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	112	100	100	100
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	124	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	87	100	100	99
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11, 616025	610275	75	100	100	100
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	42	100	100	92
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	73	100	99	93
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	85	96	92	90
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953	603604	115	100	100	100
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	51	100	99	89
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	605031	53	100	98	87
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920	300401	86	100	99	95
PLXNA3	No OMIM phenotype	300022	84	98	97	94
PLXND1	No OMIM phenotype	604282	97	100	99	96
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	66	100	100	93
PMPCA	Spinocerebellar ataxia, autosomal recessive 2, 213200	613036	133	100	100	100
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	100	100	100	98
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	61	100	99	94
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	78	100	100	100
POGZ	White-Sutton syndrome, 616364	614787	73	100	99	95
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, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450	174763	103	100	100	98
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	79	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	66	100	98	88
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	80	100	100	98
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830	614828	145	100	100	100
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094	615247	80	100	100	100
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	96	100	100	100
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	78	100	100	95
PORCN	Focal dermal hypoplasia, 305600	300651	91	100	100	97
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	68	100	100	99
POU3F3	No OMIM phenotype	602480	92	77	68	60
PPFIA4	No OMIM phenotype	603145	87	100	99	94
PPM1D	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450	605100	84	100	100	98
PPOX	Porphyria variegata, 176200	600923	103	100	100	99
PPP1CB	Noonan syndrome-like disorder with loose anagen hair 2, 617506	600590	47	100	99	89
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2, 616817	613257	80	100	100	97
PPP2CA	No OMIM phenotype	176915	59	100	99	83
PPP2R1A	Mental retardation, autosomal dominant 36, 616362	605983	124	100	100	100
PPP2R5B	No OMIM phenotype	601644	86	100	100	100
PPP2R5C	No OMIM phenotype	601645	53	98	87	75
PPP2R5D	Mental retardation, autosomal dominant 35, 616355	601646	85	100	100	96
PPP3CA	Epileptic encephalopathy, infantile or early childhood, 1, 617711	114105	65	100	96	83
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	75	100	100	97
PQBP1	Renpenning syndrome, 309500	300463	85	100	100	100
PRIM1	No OMIM phenotype	176635	43	100	92	69
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	77	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157	610087	95	100	100	98
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	96	100	96	88
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500, X-linked Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	44	100	96	81
PRR12	No OMIM phenotype	616633	101	100	96	89
PRSS12	Mental retardation, autosomal recessive 1, 249500	606709	80	100	100	98
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	122	100	100	97
PSMD12	Stankiewicz-Isidor syndrome, 617516	604450	39	96	82	60
PTCH1	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828	601309	85	100	98	94
PTCHD1	{Autism, susceptibility to, X-linked 4}, 300830	300828	69	100	100	94
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	55	100	100	93
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	103	88	79	76
PTF1A	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069	607194	127	100	100	93
PTGR1	No OMIM phenotype	601274	45	99	90	75

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	66	100	98	84
PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263	608625	70	100	100	100
PTRHD1	No OMIM phenotype	617342	109	100	100	98
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	75	100	100	90
PUF60	Verheij syndrome, 615583	604819	127	100	100	100
PUM1	Spinocerebellar ataxia 47, 617931	607204	58	100	100	95
PURA	Mental retardation, autosomal dominant 31, 616158	600473	190	100	100	100
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	86	100	100	95
PUS3	?Mental retardation, autosomal recessive 55, 617051	616283	58	100	98	93
PUS7	No OMIM phenotype	616261	49	100	96	82
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438	179035	96	100	100	96
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	109	100	100	100
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	92	100	100	98
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	80	100	100	95
RAB18	Warburg micro syndrome 3, 614222	602207	74	100	100	91
RAB27A	Griscelli syndrome, type 2, 607624	603868	44	100	98	81
RAB39B	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510	300774	52	100	100	98
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	55	100	100	93
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	57	100	98	86
RAB40AL	No OMIM phenotype	300405	208	100	100	100
RAC1	Mental retardation, autosomal dominant 48, 617751	602048	79	100	100	93
RAD21	Cornelia de Lange syndrome 4, 614701	606462	61	100	98	88
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	83	100	100	96
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	66	100	98	90
RAI1	Smith-Magenis syndrome, 182290	607642	167	100	100	100
RARB	Microphthalmia, syndromic 12, 615524	180220	70	100	100	95
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	57	100	97	85
RBBP8	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744	604124	49	100	98	81

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RBFOX1	No OMIM phenotype	605104	95	100	100	96
RBM10	TARP syndrome, 311900	300080	69	100	94	87
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	61	100	99	92
RBPJ	Adams-Oliver syndrome 3, 614814	147183	64	100	96	83
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175	607867	84	100	100	96
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	64	100	99	92
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975	605226	61	96	91	83
REV3L	No OMIM phenotype	602776	53	100	99	91
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	55	100	98	88
RHEB	No OMIM phenotype	601293	24	86	54	29
RHOBTB2	Epileptic encephalopathy, early infantile, 64, 618004	607352	133	100	100	99
RIT1	Noonan syndrome 8, 615355	609591	62	100	100	98
RLIM	Mental retardation, X-linked 61, 300978	300379	59	100	98	83
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	60	100	99	86
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	98	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	54	100	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	279	100	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	100	100	100	94
RNF113A	?Trichothiodystrophy 5, nonphotosensitive, 300953	300951	90	100	100	100
RNF125	Tenorio syndrome, 616260	610432	84	100	100	98
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	75	100	100	98
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	88	100	96	91
RORA	No OMIM phenotype	600825	71	100	100	94
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	52	98	94	81
RPL10	{Autism, susceptibility to, X-linked 5}, 300847 Mental retardation, X-linked, syndromic, 35, 300998	312173	89	100	100	100
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844	300075	40	99	85	58
RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723	616585	64	100	95	83
RTEL1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	132	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833	610436	59	100	98	88
RUBCN	?Spinocerebellar ataxia, autosomal recessive 15, 615705	613516	77	100	98	94
RUSC2	Mental retardation, autosomal recessive 61, 617773	611053	155	100	100	100
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	91	100	98	87
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480	602218	113	100	100	99
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	58	100	97	80
SATB2	Glass syndrome, 612313	608148	86	100	100	95
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	72	100	100	98
SC5D	Lathosterolosis, 607330	602286	70	100	100	98
SCAF4	No OMIM phenotype	616023	54	100	95	82
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634	182389	76	100	100	96
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	165	100	100	94
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	80	100	100	95
SCN3A	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938	182391	70	100	99	94
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	96	100	100	96
SCO1	Mitochondrial complex IV deficiency, 220110	603644	97	100	100	97
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	126	100	100	100
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	128	100	97	90
SEMA3E	?CHARGE syndrome, 214800	608166	53	100	99	87
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	65	100	99	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	56	100	95	79
SETBP1	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	87	99	98	97
SETD1A	No OMIM phenotype	611052	113	100	98	96
SETD2	Luscan-Lumish syndrome, 616831	612778	57	100	100	95
SETD5	Mental retardation, autosomal dominant 23, 615761	615743	71	100	100	96
SF1	No OMIM phenotype	601516	57	92	85	80
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	102	100	95	89
SH3KBP1	No OMIM phenotype	300374	46	99	85	60
SHANK2	{Autism susceptibility 17}, 613436	603290	119	100	99	96
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	106	98	91	84
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	116	100	100	99
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	51	100	98	89
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434	300579	57	100	99	95
SIK1	Epileptic encephalopathy, early infantile, 30, 616341	605705	125	100	100	100
SIL1	Marinesco-Sjogren syndrome, 248800	608005	94	100	100	99
SIN3A	Witteveen-Kolk syndrome, 613406	607776	59	100	99	94
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	149	100	99	95
SKI	Shprintzen-Goldberg syndrome, 182212	164780	118	100	100	99
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	56	100	98	89
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905	608305	119	100	100	97
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	66	100	100	93
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	73	100	100	94
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	61	100	100	96
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	84	100	100	95
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105	600300	58	100	100	93
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	600229	122	100	100	100
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	79	100	100	96
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	121	100	100	96

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SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	99	100	100	99
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	111	100	100	100
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	117	100	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539	603690	60	100	96	81
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	60	100	100	93
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	68	100	100	99
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	62	100	96	84
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	133	100	100	100
SLC39A12	No OMIM phenotype	608734	49	100	95	80
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721	608732	67	100	99	90
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	51	100	99	91
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	104	100	100	99
SLC6A17	Mental retardation, autosomal recessive 48, 616269	610299	108	100	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	99	100	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	91	100	99	96
SLC7A7	Lysinuric protein intolerance, 222700	603593	67	100	99	94
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243	300231	53	100	93	77
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	69	100	100	96
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	81	98	98	94
SMARCA4	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325	603254	119	100	100	100
SMARCB1	Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322 Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091	601607	121	100	100	98
SMARCC2	No OMIM phenotype	601734	67	100	99	92
SMARCE1	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174	603111	51	100	100	91

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SMC1A	Cornelia de Lange syndrome 2, 300590	300040	62	100	99	91
SMC3	Cornelia de Lange syndrome 3, 610759	606062	59	100	94	79
SMOC1	Microphthalmia with limb anomalies, 206920	608488	99	100	100	95
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	130	100	100	98
SMPD4	No OMIM phenotype	610457	108	100	100	97
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583	300105	36	95	80	59
SNAP25	?Myasthenic syndrome, congenital, 18, 616330	600322	78	100	100	92
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	142	100	100	100
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	86	100	100	100
SNRNP70	No OMIM phenotype	180740	98	100	99	97
SNRPN	Prader-Willi syndrome, 176270	182279	81	100	100	99
SNX14	Spinocerebellar ataxia, autosomal recessive 20, 616354	616105	56	100	97	83
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	120	100	97	93
SON	ZTTK syndrome, 617140	182465	77	99	95	88
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	62	100	99	89
SOS2	Noonan syndrome 9, 616559	601247	66	100	98	90
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	67	100	96	86
SOX11	Mental retardation, autosomal dominant 27, 615866	600898	160	100	100	100
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	161	100	100	100
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000	313430	66	100	96	92
SOX5	Lamb-Shaffer syndrome, 616803	604975	70	100	99	91
SPAST	Spastic paraplegia 4, autosomal dominant, 182601	604277	55	100	99	88
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577	613940	59	100	99	92
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360	610844	72	100	99	95
SPOCK1	No OMIM phenotype	602264	102	100	100	93
SPRED1	Legius syndrome, 611431	609291	49	100	96	87
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	86	100	100	97
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386	604985	116	100	100	100
SRCAP	Floating-Harbor syndrome, 136140	611421	116	100	100	99

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SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	107	100	99	95
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	48	100	98	87
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	73	100	100	98
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090	606494	71	100	99	94
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	47	99	90	77
STAG1	Mental retardation, autosomal dominant 47, 617635	604358	42	100	94	74
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	64	100	100	93
STIL	Microcephaly 7, primary, autosomal recessive, 612703	181590	56	100	99	94
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	93	100	100	100
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	91	100	100	97
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	52	100	99	95
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	79	100	99	93
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172	601485	117	100	100	96
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	69	100	100	96
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	51	100	96	81
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	82	100	100	99
SUCO	No OMIM phenotype	No id	53	100	99	90
SUMF1	Multiple sulfatase deficiency, 272200	607939	83	100	100	96
SUOX	Sulfite oxidase deficiency, 272300	606887	124	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	78	95	89	87
SUZ12	No OMIM phenotype	606245	61	100	97	90
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491	313440	55	100	99	89
SYNCRIP	No OMIM phenotype	616686	58	96	87	72
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743	608441	67	100	99	93
SYNGAP1	Mental retardation, autosomal dominant 5, 612621	603384	151	98	98	98
SYP	Mental retardation, X-linked 96, 300802	313475	67	100	100	95
SYT1	No OMIM phenotype	185605	86	100	100	97
SYT14	Spinocerebellar ataxia, autosomal recessive 11, 614229	610949	51	99	93	81
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	105	100	100	99
TAF1	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966	313650	43	100	94	75
TAF2	Mental retardation, autosomal recessive 40, 615599	604912	55	100	97	84

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TAT	Tyrosinemia, type II, 276600	613018	54	100	100	94
TBC1D20	Warburg micro syndrome 4, 615663	611663	67	100	93	93
TBC1D24	DOORS syndrome, 220500 Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	147	100	100	99
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000	612655	48	100	97	84
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	604649	99	100	99	94
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	53	99	95	82
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	616899	45	100	97	78
TBL1XR1	Mental retardation, autosomal dominant 41, 616944 Pierpont syndrome, 602342	608628	64	100	95	86
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	63	100	99	92
TBR1	No OMIM phenotype	604616	126	100	100	97
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430	602054	81	94	85	79
TCF12	Craniosynostosis 3, 615314	600480	61	100	100	95
TCF20	No OMIM phenotype	603107	95	100	100	100
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954	602272	63	100	99	93
TCF7L2	{Diabetes mellitus, type 2, susceptibility to}, 125853	602228	91	100	99	91
TCN2	Transcobalamin II deficiency, 275350	613441	107	100	100	100
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	59	100	100	94
TDP2	Spinocerebellar ataxia, autosomal recessive 23, 616949	605764	102	100	100	96
TECPR2	Spastic paraplegia 49, autosomal recessive, 615031	615000	92	100	100	99
TECR	Mental retardation, autosomal recessive 14, 614020	610057	135	100	100	99
TELO2	You-Hoover-Fong syndrome, 616954	611140	117	100	98	94
TFAP2A	Branchiooculofacial syndrome, 113620	107580	93	100	100	100
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	161	96	93	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	200	100	100	100
TGIF1	Holoprosencephaly 4, 142946	602630	138	100	100	100
TH	Segawa syndrome, recessive, 605407	191290	92	100	99	94
THOC2	Mental retardation, X-linked 12/35, 300957	300395	37	100	88	59
THOC6	Beaulieu-Boycott-Innes syndrome, 613680	615403	184	100	100	100
THRB	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650	190160	75	100	100	97
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	131	100	100	100
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130	604319	140	100	100	100
TLK2	No OMIM phenotype	608439	50	95	78	59
TM4SF20	{Specific language impairment 5}, 615432	615404	50	100	99	92
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614123	98	100	99	90
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	109	100	100	98
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	101	100	100	99
TMEM237	Joubert syndrome 14, 614424	614423	48	100	96	83
TMEM240	Spinocerebellar ataxia 21, 607454	616101	93	100	100	100
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	68	100	99	87
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	81	100	100	93
TMLHE	{Autism, susceptibility to, X-linked 6}, 300872	300777	35	100	93	65
TMPRSS7	No OMIM phenotype	No id	45	100	98	88
TMTC3	Lissencephaly 8, 617255	617218	63	100	98	89
TMX2	No OMIM phenotype	616715	61	100	93	86
TNIK	Mental retardation, autosomal recessive 54, 617028	610005	49	100	97	86
TOE1	Pontocerebellar hypoplasia, type 7, 614969	613931	89	100	99	97
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	112	100	99	96
TPO	Thyroid dyshormonogenesis 2A, 274500	606765	107	100	100	99
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	87	100	100	99
TRAIP	Seckel syndrome 9, 616777	605958	82	100	100	100

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TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	51	100	96	84
TRAPPC6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862	610397	52	100	92	71
TRAPPC9	Mental retardation, autosomal recessive 13, 613192	611966	93	100	100	97
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	217	100	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	96	100	100	100
TRIO	Mental retardation, autosomal dominant 44, 617061	601893	89	99	99	96
TRIP12	Mental retardation, autosomal dominant 49, 617752	604506	63	100	99	92
TRMT1	No OMIM phenotype	611669	96	100	99	93
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	616013	76	100	99	88
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	153	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	189	100	100	100
TSEN15	Pontocerebellar hypoplasia, type 2F, 617026	608756	66	100	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	103	99	96	96
TSHB	Hypothyroidism, congenital, nongoitrous 4, 275100	188540	79	100	100	100
TSPAN7	Mental retardation, X-linked 58, 300210	300096	47	100	98	80
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	46	100	93	72
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	50	100	98	87
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	57	100	99	87
TTI2	Mental retardation, autosomal recessive 39, 615541	614426	57	100	100	94
TUBA1A	Lissencephaly 3, 611603	602529	101	100	100	100
TUBA8	Cortical dysplasia, complex, with other brain malformations 8, 613180	605742	105	100	100	100
TUBB	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610	191130	174	100	99	96
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	242	100	99	95
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, 610031	612850	250	100	100	100
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	245	100	100	95

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TUBB4A	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	216	100	100	99
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	168	100	100	100
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335	609610	65	100	98	90
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270	610053	147	100	100	99
TUSC3	Mental retardation, autosomal recessive 7, 611093	601385	64	100	100	91
TWIST1	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746	601622	137	100	100	97
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286	606075	130	100	100	100
UBA5	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia, autosomal recessive 24, 617133	610552	41	100	87	62
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860	312180	52	100	93	75
UBE3A	Angelman syndrome, 105830	601623	54	100	100	94
UBE3B	Kaufman oculocerebrofacial syndrome, 244450	608047	110	100	100	97
UBR1	Johanson-Blizzard syndrome, 243800	605981	52	100	98	87
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801	612636	65	100	99	94
UPB1	Beta-ureidopropionase deficiency, 613161	606673	91	100	100	99
UPF3B	Mental retardation, X-linked, syndromic 14, 300676	300298	68	100	97	83
UQCC2	?Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	127	100	100	100
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	120	100	100	100
UROC1	?Urocanase deficiency, 276880	613012	107	100	100	99
USP18	Pseudo-TORCH syndrome 2, 617397	607057	120	100	95	95
USP27X	Mental retardation, X-linked 105, 300984	300975	68	100	100	95
USP7	No OMIM phenotype	602519	40	97	86	67
USP9X	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968	300072	55	100	97	89
UTRN	No OMIM phenotype	128240	66	100	99	95
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	59	100	100	93
VPS11	Leukodystrophy, hypomyelinating, 12, 616683	608549	86	100	100	97
VPS13B	Cohen syndrome, 216550	607817	63	100	98	91
VPS13C	Parkinson disease 23, autosomal recessive, early onset, 616840	608879	43	100	94	76
VPS37A	Spastic paraplegia 53, autosomal recessive, 614898	609927	39	100	97	73
VPS53	Pontocerebellar hypoplasia, type 2E, 615851	615850	75	100	99	93
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	46	100	97	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
VWA3B	?Spinocerebellar ataxia, autosomal recessive 22, 616948	614884	58	100	96	86
WAC	Desanto-Shinawi syndrome, 616708	615049	50	100	97	86
WASHC4	?Mental retardation, autosomal recessive 43, 615817	615748	59	100	96	85
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563	610657	52	100	98	86
WBP11	No OMIM phenotype	No id	44	94	67	48
WDR13	No OMIM phenotype	300512	75	100	100	98
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	62	100	99	92
WDR26	Skraban-Deardorff syndrome, 617616	617424	46	100	93	76
WDR4	No OMIM phenotype	605924	103	100	100	100
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	80	100	100	99
WDR54	No OMIM phenotype	No id	99	100	100	98
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317	613583	129	100	100	99
WDR73	Galloway-Mowat syndrome 1, 251300	616144	140	100	100	95
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, nonsyndromic, autosomal recessive 3, 617967	614218	146	100	100	100
WFS1	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296	606201	175	100	100	100
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322	605131	85	100	100	98
XIAP	Lymphoproliferative syndrome, X-linked, 2, 300635	300079	39	100	93	78
XPA	Xeroderma pigmentosum, group A, 278700	611153	62	100	99	86
XPC	Xeroderma pigmentosum, group C, 278720	613208	101	100	100	96
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	56	100	99	92
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	97	100	97	92
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	63	100	99	87
YME1L1	?Optic atrophy 11, 617302	607472	41	99	93	75
YWHAE	No OMIM phenotype	605066	66	100	100	91
YY1	Gabriele-de Vries syndrome, 617557	600013	101	100	100	100

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ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	114	100	100	100
ZBTB18	Mental retardation, autosomal dominant 22, 612337	608433	132	100	99	99
ZBTB20	Primrose syndrome, 259050	606025	144	100	100	100
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069	614064	66	100	100	98
ZC3H14	Mental retardation, autosomal recessive 56, 617125	613279	83	100	98	90
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	76	100	100	94
ZDHHC15	?Mental retardation, X-linked 91, 300577	300576	39	100	96	82
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799	300646	39	100	97	76
ZEB2	Mowat-Wilson syndrome, 235730	605802	70	100	100	99
ZFYVE26	Spastic paraplegia 15, autosomal recessive, 270700	612012	82	100	99	95
ZIC1	Craniosynostosis 6, 616602	600470	217	100	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	140	96	94	92
ZIC4	No OMIM phenotype	608948	123	100	100	99
ZMYND11	Mental retardation, autosomal dominant 30, 616083	608668	63	100	99	96
ZNF101	No OMIM phenotype	603983	50	100	99	84
ZNF292	No OMIM phenotype	616213	50	100	98	93
ZNF407	No OMIM phenotype	615894	78	100	100	99
ZNF41	No OMIM phenotype	314995	41	100	98	77
ZNF592	No OMIM phenotype	613624	123	100	100	100
ZNF674	No OMIM phenotype	300573	49	100	100	94
ZNF711	Mental retardation, X-linked 97, 300803	314990	44	100	95	74
ZNF81	No OMIM phenotype	314998	42	100	98	80
ZSWIM6	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865	615951	64	96	93	87

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "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 95 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x