Whole Exome Sequencing
Gene package Multiple congenital anomalie, version 3, 5-8-2016

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

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
A4GALT	[Blood group, P1Pk system, p phenotype], 111400 [Blood group, P1Pk system, P phenotype], 111400 NOR polyagglutination syndrome, 111400	607922	76	100	99
AAAS	Achalasia-addisonianism-alacrimia syndrome, 231550	605378	76	100	100
AAGAB	Keratoderma, palmoplantar, punctate type IA, 148600	614888	79	100	100
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	86	100	100
AARS2	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889	612035	80	100	100
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	77	100	99
ABAT	GABA-transaminase deficiency, 613163	137150	77	100	99
ABCA1	Tangier disease, 205400 HDL deficiency, type 2, 604091 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890	600046	87	100	99
ABCA12	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, autosomal recessive 4B (harlequin), 242500	607800	86	100	99
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3, 610921	601615	87	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ABCA4	Stargardt disease 1, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 {Macular degeneration, age-related, 2}, 153800 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200	601691	86	100	99
ABCB11	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479	603201	80	100	100
ABCB4	Cholestasis, progressive familial intrahepatic 3, 602347 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Gallbladder disease 1, 600803	171060	88	100	98
ABCB6	Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153	605452	79	100	100
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	51	100	98
ABCC2	Dubin-Johnson syndrome, 237500	601107	90	100	100
ABCC6	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473	603234	66	93	92
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176	600509	83	100	100
ABCC9	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850	601439	81	100	98
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	46	96	86
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	88	100	98
ABCG5	Sitosterolemia, 210250	605459	79	100	100
ABCG8	Sitosterolemia, 210250 Gallbladder disease 4, 611465	605460	91	100	100
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	62	100	94
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	100	100	100
ABL1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib	189980	97	100	100
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	98	100	100
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	88	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	607008	75	100	100
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	606885	80	100	97
ACADSB	2-methylbutyrylglycinuria, 610006	600301	80	100	95
ACADVL	VLCAD deficiency, 201475	609575	81	100	97
ACAN	Spondyloepiphyseal dysplasia, Kimberley type, 608361 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800	155760	161	100	100
ACAT1	Alpha-methylacetoacetic aciduria, 203750	607809	66	97	92
ACE	{Myocardial infarction, susceptibility to} {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 3}, 612624 [Angiotensin I-converting enzyme, benign serum increase] {SARS, progression of} Renal tubular dysgenesis, 267430 {Stroke, hemorrhagic}, 614519	106180	87	100	99
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	91	98	92
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	93	100	100
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944	171640	91	100	100
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	82	100	100
ACSL4	Mental retardation, X-linked 63, 300387	300157	44	99	91
ACSL6	Myelodysplastic syndrome Myelogenous leukemia, acute	604443	82	100	98
ACTA1	Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 ?Myopathy, scapulohumeroperoneal, 616852	102610	67	100	98
ACTA2	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042	102620	96	100	100
ACTB	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310	102630	77	100	100
ACTC1	Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Left ventricular noncompaction 4, 613424	102540	89	100	100
ACTG1	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583	102560	104	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ACTN1	Bleeding disorder, platelet-type, 15, 615193	102575	58	100	96
ACTN4	Glomerulosclerosis, focal segmental, 1, 603278	604638	76	100	98
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	86	100	100
ACVR1B	Pancreatic cancer, somatic	601300	102	100	100
ACVR2B	Heterotaxy, visceral, 4, autosomal, 613751	602730	93	100	96
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2, 600376	601284	83	100	99
ACY1	Aminoacylase 1 deficiency, 609924	104620	84	100	100
ADA	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700	608958	93	100	99
ADAM10	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590	602192	68	100	98
ADAM17	?Inflammatory skin and bowel disease, neonatal, 1, 614328	603639	75	100	95
ADAM9	Cone-rod dystrophy 9, 612775	602713	78	100	100
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	66	100	97
ADAMTS13	Thrombotic thrombocytopenic purpura, familial, 274150	604134	66	97	95
ADAMTS17	Weill-Marchesani-like syndrome, 613195	607511	82	92	89
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	83	100	98
ADAMTS2	Ehlers-Danlos syndrome, type VIIC, 225410	604539	87	99	96
ADAMTSL2	Geleophysic dysplasia 1, 231050	612277	74	98	92
ADAMTSL4	Ectopia lentis, isolated, autosomal recessive, 225100 Ectopia lentis et pupillae, 225200	610113	72	100	99
ADAR	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010	146920	96	100	100
ADAT3	Mental retardation, autosomal recessive 36, 615286	615302	53	99	95
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	600293	91	98	96
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	78	100	100
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	79	100	99
ADIPOQ	Adiponectin deficiency, 612556	605441	89	100	100
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	60	99	92
ADRB2	{Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665 Beta-2-adrenoreceptor agonist, reduced response to	109690	79	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	86	100	100
AFF2	Mental retardation, X-linked, FRAXE type, 309548	300806	45	100	93
AFG3L2	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487	604581	68	90	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
AGA	Aspartylglucosaminuria, 208400	613228	79	100	100
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	43	99	91
AGK	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691	610345	84	97	90
AGL	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400	610860	83	100	98
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	72	100	100
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121	603051	47	100	94
AGRN	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120	103320	74	99	94
AGT	{Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to} Renal tubular dysgenesis, 267430	106150	133	100	100
AGTR1	{Hypertension, essential}, 145500 Renal tubular dysgenesis, 267430	106165	93	100	100
AGXT	Hyperoxaluria, primary, type 1, 259900	604285	88	100	100
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	96	100	100
AHI1	Joubert syndrome-3, 608629	608894	77	100	98
AICDA	Immunodeficiency with hyper-IgM, type 2, 605258	605257	91	100	94
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614	300169	55	100	94
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	66	100	98
AIP	Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634 Pituitary adenoma, ACTH-secreting, 219090	605555	79	100	100
AIPL1	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393	604392	87	100	100
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300	607358	56	100	93
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	98	100	100
AK2	Reticular dysgenesis, 267500	103020	42	93	77
AKAP9	?Long QT syndrome-11, 611820	604001	57	99	90
AKR1C2	Obesity, hyperphagia, and developmental delay 46XY sex reversal 8, 614279	600450	100	100	100
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555	604741	75	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Ovarian cancer, somatic, 167000 {Schizophrenia, susceptibility to}, 181500 Proteus syndrome, somatic, 176920 Cowden syndrome 6, 615109	164730	87	100	100
AKT2	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900	164731	96	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	33	89	66
ALAD	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740	125270	73	100	94
ALAS2	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752	301300	49	100	97
ALB	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999	103600	90	100	99
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603	138250	82	100	99
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	45	98	86
ALDH2	Alcohol sensitivity, acute, 610251 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to} {Esophageal cancer, alcohol-related, susceptibility to}	100650	80	100	100
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	91	100	100
ALDH4A1	Hyperprolinemia, type II, 239510	606811	66	100	99
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	61	97	87
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	603178	80	100	100
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	76	100	99
ALDOA	Glycogen storage disease XII, 611881	103850	96	95	90
ALDOB	Fructose intolerance, 229600	612724	101	100	100
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	74	100	98
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	72	99	96
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	101	100	100
ALG13	Epileptic encephalopathy, early infantile, 36, 300884	300776	30	88	60
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	63	100	100
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	63	100	98
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	65	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ALG8	Congenital disorder of glycosylation, type Ih, 608104	608103	56	97	90
ALG9	Congenital disorder of glycosylation, type Il, 608776	606941	68	100	97
ALMS1	Alstrom syndrome, 203800	606844	105	100	100
ALOX12B	Ichthyosis, congenital, autosomal recessive 2, 242100	603741	94	100	100
ALOXE3	Ichthyosis, congenital, autosomal recessive 3, 606545	607206	86	100	100
ALPL	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300 Hypophosphatasia, adult, 146300	171760	92	100	100
ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225	606352	77	100	98
ALX1	?Frontonasal dysplasia 3, 613456	601527	96	100	100
ALX3	Frontonasal dysplasia 1, 136760	606014	65	87	80
ALX4	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451 {Craniosynostosis 5, susceptibility to}, 615529	605420	82	100	100
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	81	100	98
AMELX	Amelogenesis imperfecta, type 1E, 301200	300391	60	98	92
AMER1	Osteopathia striata with cranial sclerosis, 300373	300647	39	100	91
AMH	Persistent Mullerian duct syndrome, type I, 261550	600957	47	100	83
AMHR2	Persistent Mullerian duct syndrome, type II, 261550	600956	98	100	100
AMN	Megaloblastic anemia-1, Norwegian type, 261100	605799	58	88	82
AMPD1	Myopathy due to myoadenylate deaminase deficiency, 615511	102770	80	100	100
AMT	Glycine encephalopathy, 605899	238310	91	100	100
ANG	Amyotrophic lateral sclerosis 9, 611895	105850	70	100	100
ANGPTL3	Hypobetalipoproteinemia, familial, 2, 605019	604774	49	100	88
ANK1	Spherocytosis, type 1, 182900	612641	87	100	99
ANK2	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919	106410	85	100	100
ANKH	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600	605145	79	100	100
ANKK1	Dopamine receptor D2, reduced brain density of	608774	75	100	100
ANKRD11	KBG syndrome, 148050	611192	77	100	97
ANKRD26	Thrombocytopenia 2, 188000	610855	57	97	91
ANKS6	Nephronophthisis 16, 615382	615370	63	93	89
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728	613726	73	100	100
ANO3	Dystonia 24, 615034	610110	68	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ANO5	Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, type 2L, 611307 Miyoshi muscular dystrophy 3, 613319	608662	82	100	97
ANO6	Scott syndrome, 262890	608663	93	100	99
ANOS1	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700	300836	46	98	87
ANTXR1	{Hemangioma, capillary infantile, susceptibility to}, 602089 GAPO syndrome, 230740	606410	78	98	91
ANTXR2	Hyaline fibromatosis syndrome, 228600	608041	52	100	93
AP1S1	MEDNIK syndrome, 609313	603531	77	100	99
AP1S2	Mental retardation, X-linked syndromic 5, 304340	300629	32	97	79
AP2S1	Hypocalciuric hypercalcemia, familial, type III, 600740	602242	50	87	76
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	73	100	95
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066	607245	83	100	100
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744	607244	61	100	98
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936	602296	75	100	100
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067	607243	40	97	85
AP5Z1	Spastic paraplegia 48, autosomal recessive, 613647	613653	69	100	99
APCDD1	Hypotrichosis 1, 605389	607479	98	100	100
APOA1	ApoA-I and apoC-III deficiency, combined Hypoalphalipoproteinemia, 604091 Corneal clouding, autosomal recessive Amyloidosis, 3 or more types, 105200	107680	74	100	100
APOA2	Apolipoprotein A-II deficiency {Hypercholesterolemia, familial, modifier of}, 143890	107670	135	100	100
APOA5	{Hypertriglyceridemia, susceptibility to}, 145750 Hyperchylomicronemia, late-onset, 144650	606368	91	100	100
APOB	Hypobetalipoproteinemia, 615558 Hypercholesterolemia, due to ligand-defective apo B, 144010	107730	81	100	100
APOC2	Hyperlipoproteinemia, type Ib, 207750	608083	84	100	100
APOC3	Apolipoprotein C-III deficiency, 614028	107720	69	100	100
APRT	Adenine phosphoribosyltransferase deficiency, 614723	102600	47	100	100
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	86	100	98
AQP2	Diabetes insipidus, nephrogenic, 125800	107777	74	100	100
AQP5	Palmoplantar keratoderma, Bothnian type, 600231	600442	78	100	100
AR	Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Androgen insensitivity, partial, with or without breast cancer, 312300 {Prostate cancer, susceptibility to}, 176807 Hypospadias 1, X-linked, 300633	313700	46	96	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	86	100	100
ARG1	Argininemia, 207800	608313	71	100	100
ARHGAP26	Leukemia, juvenile myelomonocytic, somatic, 607785	605370	94	100	99
ARHGAP31	Adams-Oliver syndrome 1, 100300	610911	76	100	100
ARHGEF10	?Slowed nerve conduction velocity, AD, 608236	608136	87	100	100
ARHGEF12	No OMIM phenotype	604763	70	100	97
ARHGEF6	Mental retardation, X-linked 46, 300436	300267	50	99	88
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	47	100	87
ARID1A	Mental retardation, autosomal dominant 14, 614607	603024	83	97	94
ARID1B	Mental retardation, autosomal dominant 12, 614562	614556	83	98	93
ARL13B	Joubert syndrome 8, 612291	608922	61	100	100
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	28	80	63
ARL6	Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900 ?Retinitis pigmentosa 55, 613575	608845	48	95	81
ARMC4	Ciliary dyskinesia, primary, 23, 615451	615408	40	93	82
ARNT	No OMIM phenotype	126110	39	95	76
ARSA	Metachromatic leukodystrophy, 250100	607574	79	100	100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	611542	69	98	93
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950	300180	44	94	77
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215	300382	24	84	61
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	77	100	99
ASB10	Glaucoma 1, open angle, F, 603383	615054	61	100	100
ASCC1	Barrett esophagus/esophageal adenocarcinoma, 614266	614215	78	96	90
ASCL1	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880	100790	117	100	100
ASL	Argininosuccinic aciduria, 207900	608310	72	100	100
ASNA1	No OMIM phenotype	601913	55	100	98
ASNS	Asparagine synthetase deficiency, 615574	108370	64	93	84
ASPA	Canavan disease, 271900	608034	85	100	97
ASPM	Microcephaly 5, primary, autosomal recessive, 608716	605481	62	100	96
ASPSCR1	Alveolar soft-part sarcoma, 606243	606236	47	100	95
ASS1	Citrullinemia, 215700	603470	111	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	77	100	98
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	53	99	94
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	99	100	100
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	71	100	99
ATL1	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708	606439	68	100	100
ATL3	Neuropathy, hereditary sensory, type IF, 615632	609369	43	94	84
ATM	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic	607585	64	100	97
ATN1	Dentatorubro-pallidoluysian atrophy, 125370	607462	79	100	99
ATP13A2	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693	610513	67	100	99
ATP1A2	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481	182340	109	100	100
ATP1A3	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338	182350	96	100	99
ATP2A1	Brody myopathy, 601003	108730	89	100	100
ATP2A2	Darier disease, 124200 Acrokeratosis verruciformis, 101900	108740	95	100	100
ATP2C1	Hailey-Hailey disease, 169600	604384	70	100	98
ATP5E	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	606153	60	100	100
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	94	100	100
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive, 602722	605239	76	100	99
ATP6V1B1	Renal tubular acidosis with deafness, 267300	192132	115	100	100
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489	300011	53	99	94
ATP7B	Wilson disease, 277900	606882	90	100	100
ATP8B1	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480	602397	89	99	96
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	76	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ATR	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564	601215	81	100	98
ATRX	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580	300032	38	98	83
ATXN1	Spinocerebellar ataxia 1, 164400	601556	99	100	100
ATXN10	Spinocerebellar ataxia 10, 603516	611150	65	100	97
ATXN2	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600	601517	62	92	82
ATXN3	Machado-Joseph disease, 109150	607047	59	100	98
ATXN7	Spinocerebellar ataxia 7, 164500	607640	71	100	96
ATXN8OS	Spinocerebellar ataxia 8, 608768	603680	No coverage data		
AUH	3-methylglutaconic aciduria, type I, 250950	600529	60	100	90
AURKC	Spermatogenic failure 5, 243060	603495	64	100	100
AVP	Diabetes insipidus, neurohypophyseal, 125700	192340	38	92	72
AVPR2	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	300538	47	100	97
AXIN1	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864	603816	77	100	98
AXIN2	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500	604025	82	100	100
B2M	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200	109700	109	100	100
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	83	100	97
B3GALT6	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Ehlers-Danlos syndrome, progeroid type, 2, 615349	615291	38	75	71
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600	606374	59	94	86
B3GLCT	Peters-plus syndrome, 261540	610308	65	98	90
B3GNT2	No OMIM phenotype	605581	108	100	100
B4GALNT1	Spastic paraplegia 26, autosomal recessive, 609195	601873	86	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	70	100	100
B4GALT7	Ehlers-Danlos syndrome, progeroid type, 1, 130070	604327	87	97	95
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	76	100	99
B9D1	?Meckel syndrome 9, 614209	614144	84	100	100
B9D2	Meckel syndrome 10, 614175	611951	74	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
BAAT	Hypercholanemia, familial, 607748	602938	77	100	99
BAG3	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881	603883	91	100	100
BANF1	Nestor-Guillermo progeria syndrome, 614008	603811	60	81	61
BAP1	Tumor predisposition syndrome, 614327	603089	88	100	99
BAX	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065	600040	77	94	94
BBS1	Bardet-Biedl syndrome 1, 209900	209901	92	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	76	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	98	100	100
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	76	100	98
BBS4	Bardet-Biedl syndrome 4, 615982	600374	80	100	99
BBS5	Bardet-Biedl syndrome 5, 615983	603650	59	99	92
BBS7	Bardet-Biedl syndrome 7, 615984	607590	68	100	98
BBS9	Bardet-Biedl syndrome 9, 615986	607968	76	98	93
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	22	76	46
BCHE	Apnea, postanesthetic	177400	79	100	100
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	112	100	99
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	93	100	92
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923	614901	95	100	100
BCL10	Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300, {Sezary syndrome, somatic}, {Mesothelioma, somatic}, 156240 ?Immunodeficiency 37, 616098	603517	85	100	100
BCL2	Leukemia/lymphoma, B-cell, 2	151430	85	100	100
BCL7A	B-cell non-Hodgkin lymphoma, high-grade	601406	63	100	94
BCO1	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300	605748	94	100	97
BCOR	Microphthalmia, syndromic 2, 300166	300485	52	99	94
BCR	Leukemia, chronic myeloid, somatic, 608232 Leukemia, acute lymphocytic, somatic, 613065	151410	73	86	83
BCS1L	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358	603647	114	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
BDNF	{Memory impairment, susceptibility to} Central hypoventilation syndrome, congenital, 209880 {Obsessive-compulsive disorder, protection against}, 164230 {Bulimia nervosa, age of onset of weight loss in}, 607499 {Anorexia nervosa, susceptibility to}, 610269	113505	109	100	100
BEAN1	Spinocerebellar ataxia 31, 117210	612051	80	100	100
BEST1	Macular dystrophy, vitelliform, 2, 153700 Bestrophinopathy, autosomal recessive, 611809 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194	607854	81	100	98
BFSP1	Cataract 33, 611391	603307	69	100	98
BFSP2	Cataract 12, multiple types, 611597	603212	72	100	100
BICD2	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290	609797	68	99	93
BIN1	Myopathy, centronuclear, autosomal recessive, 255200	601248	68	100	99
BLK	Maturity-onset diabetes of the young, type 11, 613375	191305	84	100	100
BLM	Bloom syndrome, 210900	604610	74	100	99
BLNK	Agammaglobulinemia 4, 613502	604515	59	100	93
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	48	100	100
BLOC1S6	Hermansky-pudlak syndrome 9, 614171	604310	67	100	100
BLVRA	Hyperbiliverdinemia, 614156	109750	83	100	100
BMP1	Osteogenesis imperfecta, type XIII, 614856	112264	111	100	100
BMP15	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510	300247	56	100	100
BMP2	{HFE hemochromatosis, modifier of}, 235200 Brachydactyly, type A2, 112600	112261	93	100	100
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	77	100	100
BMPER	Diaphanospondylodysostosis, 608022	608699	96	100	100
BMPR1A	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900	601299	91	100	100
BMPR1B	Brachydactyly, type A2, 112600 Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849	603248	105	100	100
BMPR2	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450	600799	90	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
BOLA3	Multiple mitochondrial dysfunctions syndrome 2, 614299	613183	38	83	63
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800	613896	102	100	100
BRAF	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707	164757	60	98	88
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	69	100	97
BRIP1	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054	605882	80	100	98
BRWD3	Mental retardation, X-linked 93, 300659	300553	48	98	90
BSCL2	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924	606158	82	100	100
BSND	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522	606412	104	100	100
BTD	Biotinidase deficiency, 253260	609019	105	100	100
BTK	Agammaglobulinemia, X-linked 1, 300755 Agammaglobulinemia and isolated hormone deficiency, 307200	300300	40	100	80
BUB1	Colorectal cancer with chromosomal instability, somatic	602452	76	100	99
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	72	100	95
C10orf11	Albinism, oculocutaneous, type VII, 615179	614537	95	100	95
C10orf2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138	606075	103	100	100
C12orf57	Temtamy syndrome, 218340	615140	85	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035	613541	56	100	100
C15orf41	Dyserythropoietic anemia, congenital, type Ib, 615631	615626	44	82	69
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043	614297	71	100	100
C1GALT1C1	Tn polyagglutination syndrome, somatic, 300622	300611	44	100	99
C1QA	C1q deficiency, 613652	120550	78	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
C1QB	C1q deficiency, 613652	120570	84	100	100
C1QC	C1q deficiency, 613652	120575	106	100	100
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant, 605670	608752	68	100	96
C1S	C1s deficiency, 613783	120580	88	100	99
C2	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489	613927	53	100	96
C21orf59	Ciliary dyskinesia, primary, 26, 615500	615494	44	92	85
C2orf71	Retinitis pigmentosa 54, 613428	613425	67	100	99
C3	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378	120700	101	100	98
C4A	C4a deficiency, 614380 [Blood group, Rodgers], 614374	120810	35	95	80
C4B	C4B deficiency, 614379	120820	34	95	81
C4orf26	Amelogenesis imperfecta, type IIA4, 614832	614829	102	100	100
C5	C5 deficiency, 609536 [Eculizumab, poor response to], 615749	120900	71	100	95
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	78	100	96
C6	C6 deficiency, 612446 Combined C6/C7 deficiency	217050	99	100	100
C7	C7 deficiency, 610102	217070	88	100	98
C8A	C8 deficiency, type I, 613790	120950	89	100	100
C8B	C8 deficiency, type II, 613789	120960	94	100	98
C8orf37	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500	614477	73	100	98
C9	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591	120940	67	100	99
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	614260	52	100	99
CA12	Hyperchlorhidrosis, isolated, 143860	603263	68	100	98
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730	611492	89	100	100
CA4	Retinitis pigmentosa 17, 600852	114760	76	100	100
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	38	94	72
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	66	100	100
CABP2	Deafness, autosomal recessive 93, 614899	607314	58	100	95
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	73	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CACNA1A	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500	601011	74	100	95
CACNA1C	Timothy syndrome, 601005 Brugada syndrome 3, 611875	114205	96	100	100
CACNA1D	Sinoatrial node dysfunction and deafness, 614896 Primary aldosteronism, seizures, and neurologic abnormalities, 615474	114206	95	100	99
CACNA1F	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystrophy, X-linked, 3, 300476 Aland Island eye disease, 300600	300110	41	99	90
CACNA1S	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580	114208	89	100	99
CACNA2D4	Retinal cone dystrophy 4, 610478	608171	81	100	98
CACNB2	Brugada syndrome 4, 611876	600003	80	100	99
CACNB4	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855	601949	74	100	96
CACNG2	Mental retardation, autosomal dominant 10, 614256	602911	100	100	100
CALCOCO1	No OMIM phenotype	NA	49	98	88
CALM1	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247	114180	75	100	100
CALR	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950	109091	87	100	93
CALR3	?Cardiomyopathy, hypertrophic, 19, 613875	611414	84	100	100
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	100	100	99
CANT1	Desbuquois dysplasia 1, 251450	613165	98	100	100
CAPN3	Muscular dystrophy, limb-girdle, type 2A, 253600	114240	75	100	99
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	64	100	92
CARD11	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206	607210	93	100	99
CARD14	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200	607211	79	100	100
CARD9	Candidiasis, familial, 2, autosomal recessive, 212050	607212	75	100	100
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422	300172	39	98	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CASP10	Autoimmune lymphoproliferative syndrome, type II, 603909 Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659	601762	64	100	99
CASP8	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980	601763	75	100	99
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938	114251	90	100	100
CASR	Hypocalciuric hypercalcemia, type I, 145980 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypercalciuric hypercalcemia {Calcium, serum level of}	601199	111	100	100
CAT	Acatalasemia, 614097	115500	88	100	100
CATSPER1	Spermatogenic failure 7, 612997	606389	90	100	100
CAV1	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721	601047	78	100	100
CAV3	Muscular dystrophy, limb-girdle, type IC, 607801 Rippling muscle disease, 606072 Creatine phosphokinase, elevated serum, 123320 Myopathy, distal, Tateyama type, 614321 Cardiomyopathy, familial hypertrophic, 192600 Long QT syndrome 9, 611818	601253	144	100	100
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	68	100	100
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	82	100	99
CBX2	?46XY sex reversal 5, 613080	602770	63	100	97
CC2D1A	Mental retardation, autosomal recessive 3, 608443	610055	74	100	99
CC2D2A	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360	612013	82	100	99
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	82	100	96
CCDC103	Ciliary dyskinesia, primary, 17, 614679	614677	95	100	100
CCDC114	Ciliary dyskinesia, primary, 20, 615067	615038	59	100	94
CCDC39	Ciliary dyskinesia, primary, 14, 613807	613798	62	100	96
CCDC40	Ciliary dyskinesia, primary, 15, 613808	613799	83	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CCDC50	?Deafness, autosomal dominant 44, 607453	611051	69	100	100
CCDC65	Ciliary dyskinesia, primary, 27, 615504	611088	29	94	71
CCDC78	Myopathy, centronuclear, 4, 614807	614666	64	100	100
CCDC8	3-M syndrome 3, 614205	614145	104	100	100
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053	611204	77	100	100
CCM2	Cerebral cavernous malformations-2, 603284	607929	89	100	98
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840	610150	85	99	96
CD151	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620	602243	74	100	100
CD19	Immunodeficiency, common variable, 3, 613493	107265	82	100	100
CD247	?Immunodeficiency 25, 610163	186780	61	100	100
CD27	Lymphoproliferative syndrome 2, 615122	186711	69	100	98
CD2AP	Glomerulosclerosis, focal segmental, 3, 607832	604241	65	99	89
CD320	Methylmalonic aciduria due to transcobalamin receptor defect, 613646	606475	60	100	95
CD36	[Macrothrombocytopenia] Platelet glycoprotein IV deficiency, 608404 {Malaria, cerebral, susceptibility to}, 611162 {Malaria, cerebral, reduced risk of}, 611162 {Coronary heart disease, susceptibility to, 7}, 610938	173510	75	100	100
CD3D	Immunodeficiency 19, 615617	186790	92	100	92
CD3E	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615	186830	68	100	96
CD3G	Immunodeficiency 17, CD3 gamma deficient, 615607	186740	93	100	100
CD4	OKT4 epitope deficiency, 613949	186940	83	100	100
CD40	Immunodeficiency with hyper-IgM, type 3, 606843	109535	91	100	100
CD40LG	Immunodeficiency, X-linked, with hyper-IgM, 308230	300386	57	96	87
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300	107271	103	100	100
CD79A	Agammaglobulinemia 3, 613501	112205	85	100	100
CD79B	Agammaglobulinemia 6, 612692	147245	125	100	100
CD81	Immunodeficiency, common variable, 6, 613496	186845	98	100	100
CD8A	CD8 deficiency, familial, 608957	186910	67	100	98
CD96	C syndrome, 211750	606037	81	100	97
CDAN1	Dyserythropoietic anemia, congenital, type Ia, 224120	607465	73	100	99
CDC6	Meier-Gorlin syndrome 5, 613805	602627	72	100	100
CDC73	Hyperparathyroidism-jaw tumor syndrome, 145001 Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266	607393	68	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CDH1	Endometrial carcinoma, somatic, 608089 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 {Prostate cancer, susceptibility to}, 176807	192090	95	100	100
CDH15	Mental retardation, autosomal dominant 3, 612580	114019	84	100	97
CDH23	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067	605516	119	100	100
CDH3	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280	114021	94	100	100
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	103	98	98
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804	608201	69	100	97
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	47	98	94
CDKN1B	Multiple endocrine neoplasia, type IV, 610755	600778	73	100	100
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	46	80	72
CDKN2A	{Melanoma, cutaneous malignant, 2}, 155601 Melanoma and neural system tumor syndrome, 155755 Pancreatic cancer/melanoma syndrome, 606719 Orolaryngeal cancer, multiple,	600160	50	100	100
CDON	Holoprosencephaly 11, 614226	608707	90	100	100
CDSN	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300	602593	62	100	97
CDT1	Meier-Gorlin syndrome 4, 613804	605525	65	100	97
CEACAM16	?Deafness, autosomal dominant 4B, 614614	614591	88	100	100
CEBPA	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626	116897	65	91	61
CEBPE	Specific granule deficiency, 245480	600749	57	100	98
CECR1	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	34	93	73
CEL	Maturity-onset diabetes of the young, type VIII, 609812	114840	90	94	86
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676	609279	80	100	98
CEP135	?Microcephaly 8, primary, autosomal recessive, 614673	611423	60	100	97
CEP152	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823	613529	76	100	98
CEP164	Nephronophthisis 15, 614845	614848	68	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CEP19	Morbid obesity and spermatogenic failure, 615703	615586	72	100	100
CEP290	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 ?Bardet-Biedl syndrome 14, 615991	610142	55	98	92
CEP41	Joubert syndrome 15, 614464	610523	64	100	93
CEP57	Mosaic variegated aneuploidy syndrome 2, 614114	607951	60	100	99
CERKL	Retinitis pigmentosa 26, 608380	608381	73	100	97
CERS3	Ichthyosis, congenital, autosomal recessive 9, 615023	615276	39	98	81
CES1	Carboxylesterase 1 deficiency	114835	68	98	90
CETP	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470	118470	88	100	100
CFAP53	Heterotaxy, visceral, 6, autosomal recessive, 614779	614759	87	100	99
CFC1	Heterotaxy, visceral, 2, autosomal, 605376	605194	35	76	55
CFD	Complement factor D deficiency, 613912	134350	55	100	94
CFH	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Complement factor H deficiency, 609814 {Macular degeneration, age-related, 4}, 610698 Basal laminar drusen, 126700	134370	80	99	96
CFHR5	Nephropathy due to CFHR5 deficiency, 614809	608593	64	100	99
CFI	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439	217030	71	99	96
CFL2	Nemaline myopathy 7, autosomal recessive, 610687	601443	67	100	93
CFP	Properdin deficiency, X-linked, 312060	300383	48	100	97
CFTR	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF {Pancreatitis, idiopathic}, 167800 {Hypertrypsinemia, neonatal} {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400	602421	88	100	98
CHAT	Myasthenic syndrome, congenital, 6, presynaptic, 254210	118490	85	92	87
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	75	100	98
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	80	100	98
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	57	100	99
CHM	Choroideremia, 303100	300390	48	100	94
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	80	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CHMP2B	Dementia, familial, nonspecific, 600795 Amyotrophic lateral sclerosis 17, 614696	609512	60	100	97
CHMP4B	Cataract 31, multiple types, 605387	610897	65	99	91
CHN1	Duane retraction syndrome 2, 604356	118423	87	100	98
CHRDL1	Megalocornea 1, X-linked 309300	300350	50	100	96
CHRM3	?Prune belly syndrome, 100100	118494	73	100	100
CHRNA1	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290	100690	89	100	100
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353	118502	119	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	91	97	94
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314	100710	102	100	100
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375	118507	102	100	97
CHRND	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290	100720	93	100	100
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931	100725	74	100	100
CHRNG	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290	100730	96	100	100
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1, 601776	608429	84	97	94
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	603799	61	100	94
CHST6	Macular corneal dystrophy, 217800	605294	141	100	100
CHSY1	Temtamy preaxial brachydactyly syndrome, 605282	608183	83	97	95
CHUK	Cocoon syndrome, 613630	600664	71	100	95
CIB2	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869	605564	109	100	100
CIITA	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300	600005	75	100	99
CISD2	Wolfram syndrome 2, 604928	611507	98	100	100
CITED2	Ventricular septal defect 2, 614431 Atrial septal defect 8, 614433	602937	102	100	100
CLCF1	Cold-induced sweating syndrome 2, 610313	607672	45	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CLCN1	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive	118425	94	100	100
CLCN2	{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 Leukoencephalopathy with ataxia, 615651	600570	73	100	99
CLCN5	Dent disease, 300009 Nephrolithiasis, type I, 310468 Hypophosphatemic rickets, 300554 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990	300008	43	100	94
CLCN7	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600	602727	86	100	99
CLCNKA	Bartter syndrome, type 4b, digenic, 613090	602024	74	98	91
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	70	98	86
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626	603718	96	100	100
CLDN14	Deafness, autosomal recessive 29, 614035	605608	55	100	100
CLDN16	Hypomagnesemia 3, renal, 248250	603959	92	100	100
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	79	100	97
CLEC7A	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079	606264	74	100	100
CLIC2	?Mental retardation, X-linked, syndromic 32, 300886	300138	46	100	100
CLMP	Congenital short bowel syndrome, 615237	611693	62	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	68	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	73	100	94
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	72	100	91
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	100	100	100
CLPP	Perrault syndrome 3, 614129	601119	60	97	90
CLRN1	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180	606397	105	100	100
CNBP	Myotonic dystrophy 2, 602668	116955	78	100	100
CNGA1	Retinitis pigmentosa 49, 613756	123825	62	91	91
CNGA3	Achromatopsia-2, 216900	600053	98	100	100
CNGB1	Retinitis pigmentosa 45, 613767	600724	81	100	98
CNGB3	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200	605080	80	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	113	100	100
CNNM4	Jalili syndrome, 217080	607805	129	99	98
CNTN1	?Myopathy, congenital, Compton-North, 612540	600016	80	100	100
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042	604569	85	100	100
CNTNAP4	No OMIM phenotype	610518	86	100	99
COA5	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500	613920	94	100	100
COASY	Neurodegeneration with brain iron accumulation 6, 615643	609855	74	100	100
COCH	Deafness, autosomal dominant 9, 601369	603196	111	100	100
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	91	100	98
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	71	100	99
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	60	100	95
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	72	99	96
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	79	100	100
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	72	100	100
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500	120110	66	100	97
COL11A1	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520	120280	56	98	88
COL11A2	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524	120290	64	100	97
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650 Epithelial recurrent erosion dystrophy, 122400	113811	68	98	95
COL18A1	Knobloch syndrome, type 1, 267750	120328	71	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
COL1A1	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 Caffey disease, 114000 {Bone mineral density variation QTL, osteoporosis}, 166710	120150	76	99	94
COL1A2	Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type II, 166210 {Osteoporosis, postmenopausal}, 166710 Ehlers-Danlos syndrome, cardiac valvular form, 225320	120160	71	100	96
COL2A1	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Spondyloperipheral dysplasia, 271700 Osteoarthritis with mild chondrodysplasia, 604864 Vitreoretinopathy with phalangeal epiphyseal dysplasia Platyspondylic skeletal dysplasia, Torrance type, 151210 Otospondylomegaepiphyseal dysplasia, 215150 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600 Stickler sydrome, type I, nonsyndromic ocular, 609508 Czech dysplasia, 609162 Spondyloepiphyseal dysplasia, Stanescu type, 616583	120140	74	100	99
COL3A1	Ehlers-Danlos syndrome, type IV, 130050	120180	63	99	93
COL4A1	Porencephaly 1, 175780 Brain small vessel disease with or without ocular anomalies, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000	120130	76	99	96
COL4A2	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519	120090	69	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
COL4A3	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200	120070	62	98	97
COL4A4	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign	120131	62	100	96
COL4A5	Alport syndrome, 301050	303630	34	97	79
COL5A1	Ehlers-Danlos syndrome, classic type, 130000	120215	77	99	97
COL5A2	Ehlers-Danlos syndrome, classic type, 130000	120190	68	100	97
COL6A1	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090	120220	90	100	100
COL6A2	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600	120240	94	100	99
COL6A3	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411	120250	93	100	99
COL7A1	Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa, pretibial, 131850 EBD, Bart type, 132000 EBD, localisata variant Transient bullous of the newborn, 131705 Epidermolysis bullosa pruriginosa, 604129 Toenail dystrophy, isolated, 607523 EBD inversa, 226600	120120	78	100	99
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	43	95	82
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	71	99	95
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 ?Stickler syndrome, type V, 614284	120260	50	100	94
COL9A3	Epiphyseal dysplasia, multiple, 3, 600969 Epiphyseal dysplasia, multiple, with myopathy {Intervertebral disc disease, susceptibility to}, 603932	120270	61	99	94
COLEC11	3MC syndrome 2, 265050	612502	99	100	100
COLQ	Myasthenic syndrome, congenital, 5, 603034	603033	81	100	99
COMP	Pseudoachondroplasia, 177170 Epiphyseal dysplasia, multiple, 1, 132400	600310	84	92	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	94
COQ6	Coenzyme Q10 deficiency, primary, 6, 614650	614647	78	100	98
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	No coverage data		
COQ8B	Nephrotic syndrome, type 9, 615573	615567	No coverage data		
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	75	100	95
CORIN	Preeclampsia/eclampsia 5, 614595	605236	83	100	100
CORO1A	Immunodeficiency 8, 615401	605000	86	99	93
COX10	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000	602125	105	100	100
COX14	?Mitochondrial complex IV deficiency, 220110	614478	59	100	100
COX15	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119	603646	86	100	100
COX20	Mitochondrial complex IV deficiency, 220110	614698	14	60	25
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714	607976	77	100	100
COX7B	Linear skin defects with multiple congenital anomalies, 300887	300885	33	100	83
CP	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290	117700	50	87	77
CPA6	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418	609562	104	100	100
CPN1	Carboxypeptidase N deficiency, 212070	603103	61	100	96
CPOX	Coproporphyria, 121300 Harderoporphyria, 121300	612732	79	100	99
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	78	100	99
CPT1A	CPT deficiency, hepatic, type IA, 255120	600528	112	100	100
CPT2	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212	600650	120	99	96
CR2	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699	120650	87	100	99
CRADD	Mental retardation, autosomal recessive 34, 614499	603454	73	100	96
CRB1	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870	604210	84	100	100
CRBN	Mental retardation, autosomal recessive 2, 607417	609262	87	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CREB1	Histiocytoma, angiomatoid fibrous, somatic, 612160	123810	85	100	97
CREBBP	Rubinstein-Taybi syndrome, 180849	600140	81	100	97
CRELD1	{Atrioventricular septal defect, susceptibility to, 2}, 606217 Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217	607170	75	100	97
CRLF1	Cold-induced sweating syndrome 1, 272430	604237	84	91	91
CRTAP	Osteogenesis imperfecta, type VII, 610682	605497	92	100	100
CRTC1	Mucoepidermoid salivary gland carcinoma	607536	63	98	91
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	104	100	100
CRYAA	Cataract 9, multiple types, 604219	123580	126	100	100
CRYAB	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184	123590	77	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	93	100	98
CRYBA4	Cataract 23, 610425	123631	67	100	93
CRYBB1	Cataract 17, multiple types, 611544	600929	87	100	100
CRYBB2	Cataract 3, multiple types, 601547	123620	92	100	100
CRYBB3	Cataract 22, autosomal recessive, 609741	123630	92	100	100
CRYGB	Cataract 39, multiple types, autosomal dominant, 615188	123670	82	100	100
CRYGC	Cataract 2, multiple types, 604307	123680	85	100	100
CRYGD	Cataract 4, multiple types, 115700	123690	63	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	74	100	93
CRYM	Deafness, autosomal dominant 40, 616357	123740	70	100	100
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	164770	81	100	99
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4, 300770	306250	64	92	89
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5, 614370	138981	69	100	98
CSF3R	?Neutrophilia, hereditary, 162830	138971	64	100	99
CSNK1D	Advanced sleep-phase syndrome, familial, 2, 615224	600864	104	100	96
CSPP1	Joubert syndrome 21, 615636	611654	38	96	81
CSRP3	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124	600824	76	100	100
CST3	Cerebral amyloid angiopathy, 105150 Macular degeneration, age-related, 11, 611953	604312	49	100	95
CSTA	Peeling skin syndrome 4, 607936	184600	67	100	100
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	124	100	97
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	71	100	100
CTCF	Mental retardation, autosomal dominant 21, 615502	604167	47	96	89
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	74	95	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CTH	Cystathioninuria, 219500 Homocysteine, total plasma, elevated	607657	93	100	100
CTHRC1	Barrett esophagus/esophageal adenocarcinoma, 614266	610635	70	100	95
CTNNA3	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616	607667	88	100	98
CTNNB1	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Pilomatricoma, somatic, 132600 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550	116806	81	100	100
CTNS	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800	606272	99	100	100
CTSA	Galactosialidosis, 256540	613111	81	100	100
CTSC	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650	602365	72	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	83	100	96
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	43	82	70
CTSK	Pycnodysostosis, 265800	601105	96	100	100
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	83	99	98
CUL3	Pseudohypoaldosteronism, type IIE, 614496	603136	61	100	97
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354	300304	39	100	92
CUL7	3-M syndrome 1, 273750	609577	78	100	98
CXCR4	WHIM syndrome, 193670 Myelokathexis, isolated	162643	86	100	100
CYB5A	Methemoglobinemia, type IV, 250790	613218	88	100	100
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	86	100	100
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690	608508	52	88	78
CYBB	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645	300481	54	100	100
CYC1	Mitochondrial complex III deficiency, nuclear type 6, 615453	123980	65	100	90
CYCS	Thrombocytopenia 4, 612004	123970	25	95	65
CYLD	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606	605018	80	100	99
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743	118485	77	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900	610613	113	100	98
CYP11B2	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 {Low renin hypertension, susceptibility to} Aldosterone to renin ratio raised	124080	118	100	99
CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110	609300	77	100	100
CYP19A1	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300	107910	96	100	100
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229	601771	73	100	100
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910	613815	71	100	100
CYP24A1	Hypercalcemia, infantile, 143880	126065	100	100	100
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	605207	101	100	100
CYP26C1	Focal facial dermal dysplasia 4, 614974	608428	60	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	95	99	95
CYP27B1	Vitamin D-dependent rickets, type I, 264700	609506	89	100	98
CYP2A6	Coumarin resistance, 122700 {Nicotine addiction, protection from}, 188890 {Lung cancer, resistance to}, 211980	122720	73	100	96
CYP2B6	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546	123930	86	100	97
CYP2C19	Mephenytoin poor metabolizer, 609535 Omeprazole poor metabolizer, 609535 Proguanil poor metabolizer, 609535 Clopidogrel, impaired responsiveness to, 609535	124020	124	100	100
CYP2C8	Rhabdomyolysis, cerivastatin-induced	601129	96	100	100
CYP2C9	Tolbutamide poor metabolizer Warfarin sensitivity, 122700	601130	136	100	100
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation, 600081	608713	74	100	100
CYP2U1	Spastic paraplegia 56, autosomal recessive, 615030	610670	89	96	93
CYP46A1	No OMIM phenotype	604087	35	89	68
CYP4F22	Ichthyosis, congenital, autosomal recessive 5, 604777	611495	82	100	100
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	89	100	100
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800	603711	56	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	91	100	100
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538	128239	108	100	100
DARS	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281	603084	40	93	77
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	71	100	97
DBH	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360	609312	103	100	100
DBT	Maple syrup urine disease, type II, 248600	248610	70	100	95
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	71	100	98
DCC	Mirror movements 1, 157600 Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239	120470	85	100	97
DCHS1	Van Maldergem syndrome 1, 601390 Mitral valve prolapse 2, 607829	603057	70	99	97
DCLRE1C	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554	605988	69	97	93
DCN	Corneal dystrophy, congenital stromal, 610048	125255	98	100	100
DCTN1	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605	601143	84	100	98
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067	300121	47	98	85
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740	600811	98	100	100
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	95	100	100
DDHD1	Spastic paraplegia 28, autosomal recessive, 609340	614603	84	98	95
DDHD2	Spastic paraplegia 54, autosomal recessive, 615033	615003	86	100	100
DDOST	?Congenital disorder of glycosylation, type Ir, 614507	602202	79	100	99
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	191311	96	100	100
DDX11	Warsaw breakage syndrome, 613398	601150	39	78	66
DDX59	Orofaciodigital syndrome V, 174300	615464	59	99	89
DEPDC5	Epilepsy, familial focal, with variable foci, 604364	614191	53	99	90
DES	Myopathy, myofibrillar, 1, 601419 Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325	125660	92	100	100
DFNA5	Deafness, autosomal dominant 5, 600994	608798	88	100	100
DFNB59	Deafness, autosomal recessive 59, 610220	610219	79	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DGKE	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008	601440	75	100	94
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880	601465	88	100	100
DHCR24	Desmosterolosis, 602398	606418	104	100	100
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	114	100	100
DHDDS	Retinitis pigmentosa 59, 613861	608172	73	100	100
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	21	74	44
DHH	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420	605423	83	100	100
DHODH	Miller syndrome, 263750	126064	97	100	100
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	100	100	100
DIABLO	Deafness, autosomal dominant 64, 614152	605219	107	100	100
DIAPH1	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	80	100	96
DIAPH2	Premature ovarian failure, 300511	300108	33	98	76
DIAPH3	Auditory neuropathy, autosomal dominant, 1, 609129	614567	54	100	92
DICER1	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Rhabdomyosarcoma, embryonal, 2, 180295	606241	82	99	98
DIP2B	Mental retardation, FRA12A type, 136630	611379	83	100	98
DIS3L2	Perlman syndrome, 267000	614184	99	100	98
DKC1	Dyskeratosis congenita, X-linked, 305000	300126	57	100	94
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	62	100	95
DLC1	Colorectal cancer, somatic, 114500	604258	97	100	100
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	84	100	97
DLG3	Mental retardation, X-linked 90, 300850	300189	40	100	84
DLL3	Spondylocostal dysostosis 1, autosomal recessive, 277300	602768	56	92	82
DLX3	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510	600525	96	100	100
DMD	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045	300377	50	99	93
DMGDH	Dimethylglycine dehydrogenase deficiency, 605850	605849	96	100	97
DMP1	Hypophosphatemic rickets, AR, 241520	600980	62	100	99
DMPK	Myotonic dystrophy 1, 160900	605377	70	100	97
DNA2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807	601810	45	95	80

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DNAAF1	Ciliary dyskinesia, primary, 13, 613193	613190	88	100	100
DNAAF2	Ciliary dyskinesia, primary, 10, 612518	612517	70	100	97
DNAAF3	Ciliary dyskinesia, primary, 2, 606763	614566	70	100	97
DNAAF5	Ciliary dyskinesia, primary, 18, 614874	614864	82	90	82
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	603339	87	100	99
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	603335	85	100	99
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	604366	84	100	97
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	605483	107	100	98
DNAJB2	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 ?Charcot-Marie-Tooth disease, axonal, type 2T, 616233	604139	75	100	94
DNAJB6	Muscular dystrophy, limb-girdle, type 1E, 603511	611332	26	80	53
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	35	88	60
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	111	100	100
DNAJC6	Parkinson disease 19, juvenile-onset, 615528	608375	88	100	100
DNAL1	Ciliary dyskinesia, primary, 16, 614017	610062	57	90	89
DNASE1L3	Systemic lupus erythematosus 16, 614420	602244	89	100	100
DNM1L	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388	603850	62	100	96
DNM2	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Lethal congenital contracture syndrome 5, 615368	602378	83	100	100
DNMT1	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121	126375	70	100	98
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	89	100	99
DOCK6	Adams-Oliver syndrome 2, 614219	614194	73	99	98
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700	611432	84	100	99
DOK7	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence, 208150	610285	50	94	88
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	84	100	100
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	80	100	99
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	69	100	100
DPM2	Congenital disorder of glycosylation, type Iu, 615042	603564	55	100	100
DPM3	Congenital disorder of glycosylation, type Io, 612937	605951	100	100	100
DPP6	{Ventricular fibrillation, paroxysmal familial, 2}, 612956 Mental retardation, autosomal dominant 33, 616311	126141	70	99	94
DPY19L2	Spermatogenic failure 9, 613958	613893	32	62	53
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	79	98	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DPYS	Dihydropyrimidinuria, 222748	613326	103	100	98
DRC1	Ciliary dyskinesia, primary, 21, 615294	615288	34	96	79
DRD2	No OMIM phenotype	126450	84	100	100
DRD4	Autonomic nervous system dysfunction [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465	126452	70	97	82
DRD5	{Blepharospasm, primary benign}, 606798 Dystonia, primary cervical {Attention deficit-hyperactivity disorder, susceptibility to}, 143465	126453	62	100	100
DSC2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476	125645	73	100	98
DSC3	?Hypotrichosis and recurrent skin vesicles, 613102	600271	61	100	97
DSG1	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508	125670	74	100	99
DSG2	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877	125671	71	100	100
DSG4	Hypotrichosis 6, 607903	607892	87	100	98
DSP	Keratosis palmoplantaris striata II, 612908 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821	125647	78	100	99
DSPP	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420	125485	49	84	61
DST	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425	113810	90	100	98
DTNA	Left ventricular noncompaction 1, with or without congenital heart defects, 604169	601239	84	100	98
DTNBP1	{Schizophrenia}, 181500 Hermansky-Pudlak syndrome 7, 614076	607145	73	100	97
DUOX2	Thryoid dyshormonogenesis 6, 607200	606759	87	95	93
DUOXA2	Thyroid dyshormonogenesis 5, 274900	612772	70	100	100
DUSP6	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269	602748	67	100	98
DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	64	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	86	100	99
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	603297	64	99	94
DYRK1A	Mental retardation, autosomal dominant 7, 614104	600855	99	100	100
DYSF	Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768 Miyoshi muscular dystrophy 1, 254130	603009	97	100	100
DYX1C1	{Dyslexia, susceptibility to, 1}, 127700 Ciliary dyskinesia, primary, 25, 615482	608706	63	100	96
EARS2	Combined oxidative phosphorylation deficiency 12, 614924	612799	71	100	100
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960	300205	43	100	93
ECE1	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500	600423	97	98	97
ECEL1	Arthrogryposis, distal, type 5D, 615065	605896	47	92	78
ECM1	Urbach-Wiethe disease, 247100	602201	100	100	100
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500	300451	39	98	92
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630	604095	84	100	100
EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940	606603	63	100	95
EDN1	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}	131240	72	100	100
EDN3	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712	131242	103	100	100
EDNRA	{Migraine, resistance to}, 157300 Mandibulofacial dysostosis with alopecia, 616367	131243	96	100	100
EDNRB	{Hirschsprung disease, susceptibility to, 2}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580	131244	93	100	100
EFEMP1	Doyne honeycomb degeneration of retina, 126600	601548	108	100	100
EFEMP2	Cutis laxa, autosomal recessive, type IB, 614437	604633	82	100	100
EFNB1	Craniofrontonasal dysplasia, 304110	300035	58	100	100
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536	603892	84	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
EGF	Hypomagnesemia 4, renal, 611718	131530	80	100	100
EGFR	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980 ?Inflammatory skin and bowel disease, neonatal, 2, 616069	131550	89	100	100
EGLN1	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070	606425	63	92	73
EGR2	Neuropathy, congenital hypomyelinating, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900	129010	61	100	95
EHMT1	Kleefstra syndrome, 610253	607001	97	99	99
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	73	98	91
EIF2AK4	Pulmonary venoocclusive disease 2, 234810	609280	52	98	88
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896	606686	89	100	100
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606454	85	100	100
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896	606273	100	100	100
EIF2B4	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606687	74	100	96
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	603945	88	100	100
EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305	608546	35	99	81
EIF4G1	{Parkinson disease 18}, 614251	600495	92	100	99
ELAC2	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440	605367	73	100	100
ELANE	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700	130130	81	100	98
ELN	Supravalvar aortic stenosis, 185500 Cutis laxa, AD, 123700	130160	63	100	99
ELOVL4	Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190	605512	57	100	98
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300	300384	62	100	96
EMG1	Bowen-Conradi syndrome, 211180	611531	78	100	100
EMX2	Schizencephaly, 269160	600035	95	100	100
ENAM	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650	606585	74	100	100
ENG	Telangiectasia, hereditary hemorrhagic, type 1, 187300	131195	87	100	99
ENO3	?Glycogen storage disease XIII, 612932	131370	89	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ENPP1	{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522	173335	79	97	91
ENTPD1	Spastic paraplegia 64, autosomal recessive, 615683	601752	88	100	100
EOGT	Adams-Oliver syndrome 4, 615297	614789	45	96	87
EP300	Rubinstein-Taybi syndrome 2, 613684 Colorectal cancer, somatic, 114500	602700	90	100	100
EPAS1	Erythrocytosis, familial, 4, 611783	603349	89	100	100
EPB41	Elliptocytosis-1, 611804	130500	86	100	99
EPB42	Spherocytosis, type 5, 612690	177070	102	100	100
EPCAM	Diarrhea 5, with tufting enteropathy, congenital, 613217 Colorectal cancer, hereditary nonpolyposis, type 8, 613244	185535	42	100	97
EPG5	Vici syndrome, 242840	615068	42	96	83
EPHA2	Cataract 6, multiple types, 116600	176946	83	100	99
EPHB2	{Prostate cancer/brain cancer susceptibility, somatic}, 603688	600997	110	98	98
EPHX1	?Fetal hydantoin syndrome Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 {Preeclampsia, susceptibility to}, 189800	132810	99	100	100
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	66	87	83
EPX	[Eosinophil peroxidase deficiency], 261500	131399	101	100	100
ERBB2	Adenocarcinoma of lung, somatic, 211980 Glioblastoma, somatic, 137800 Gastric cancer, somatic, 613659 Ovarian cancer, somatic,	164870	96	98	98
ERBB3	Lethal congenital contractural syndrome 2, 607598	190151	84	100	100
ERBB4	Amyotrophic lateral sclerosis 19, 615515	600543	95	100	99
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	73	100	92
ERCC2	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 Cerebrooculofacioskeletal syndrome 2, 610756	126340	77	100	100
ERCC3	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390	133510	81	100	100
ERCC4	Xeroderma pigmentosum, group F, 278760 ?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760	133520	80	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ERCC5	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Cerebrooculofacioskeletal syndrome 3, 616570	133530	73	100	99
ERCC6	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980	609413	93	100	100
ERCC6L2	Bone marrow failure syndrome 2, 615715	615667	48	97	88
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	60	100	96
ERF	Craniosynostosis 4, 600775	611888	56	100	87
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225	611605	92	100	100
ESCO2	Roberts syndrome, 268300 SC phocomelia syndrome, 269000	609353	58	98	92
ESPN	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant	606351	37	84	66
ESR1	Estrogen resistance, 615363 {HDL response to hormone replacement, augmented} {Migraine, susceptibility to}, 157300 {Atherosclerosis, susceptibility to} {Myocardial infarction, susceptibility to}, 608446 {Breast cancer}, 114480	133430	75	100	99
ESRRB	Deafness, autosomal recessive 35, 608565	602167	80	100	100
ETFA	Glutaric acidemia IIA, 231680	608053	54	100	97
ETFB	Glutaric acidemia IIB, 231680	130410	82	100	100
ETFDH	Glutaric acidemia IIC, 231680	231675	75	100	97
ETHE1	Ethylmalonic encephalopathy, 602473	608451	67	100	99
ETV6	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216	600618	79	100	100
EVC	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530	604831	77	94	91
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	607261	78	97	95
EWSR1	Ewing sarcoma, 612219 Neuroepithelioma, 612219	133450	76	100	96
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	81	100	100
EXPH5	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028	612878	74	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
EXT1	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300	608177	75	100	100
EXT2	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	91	100	100
EYA1	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 ?Otofaciocervical syndrome, 166780	601653	78	100	96
EYA4	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362	603550	82	100	100
EYS	Retinitis pigmentosa 25, 602772	612424	76	100	98
EZH2	Weaver syndrome, 277590	601573	84	100	99
F10	Factor X deficiency, 227600	613872	95	100	100
F11	Factor XI deficiency, autosomal recessive, 612416 Factor XI deficiency, autosomal dominant, 612416	264900	103	100	99
F12	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618	610619	77	100	100
F13A1	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050	134570	96	100	100
F13B	Factor XIIIB deficiency, 613235	134580	67	100	96
F2	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Stroke, ischemic, susceptibility to}, 601367 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390	176930	78	100	98
F5	Factor V deficiency, 227400 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 {Stroke, ischemic, susceptibility to}, 601367 {Budd-Chiari syndrome}, 600880 Thrombophilia due to activated protein C resistance, 188055 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389	612309	103	100	100
F7	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446	613878	87	100	100
F8	Hemophilia A, 306700	300841	48	100	94
F9	Hemophilia B, 306900 {Warfarin sensitivity}, 122700 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807	300746	51	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FA2H	Spastic paraplegia 35, autosomal recessive, 612319	611026	55	98	87
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759	602457	99	100	100
FAH	Tyrosinemia, type I, 276700	613871	112	100	100
FAM111A	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361	615292	75	99	96
FAM111B	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704	615584	61	99	94
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	67	100	94
FAM134B	Neuropathy, hereditary sensory and autonomic, type IIB, 613115	613114	62	97	91
FAM161A	Retinitis pigmentosa 28, 606068	613596	70	100	100
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690	611062	67	100	94
FAM20C	Raine syndrome, 259775	611061	79	100	99
FAM58A	STAR syndrome, 300707	300708	32	76	64
FAM83H	Amelogenesis imperfecta, type III, 130900	611927	64	99	92
FAN1	Interstitial nephritis, karyomegalic, 614817	613534	77	100	100
FANCA	Fanconi anemia, complementation group A, 227650	607139	75	99	95
FANCB	Fanconi anemia, complementation group B, 300514	300515	35	99	86
FANCC	Fanconi anemia, complementation group C, 227645	613899	59	100	98
FANCD2	Fanconi anemia, complementation group D2, 227646	613984	67	96	91
FANCE	Fanconi anemia, complementation group E, 600901	613976	63	89	85
FANCF	Fanconi anemia, complementation group F, 603467	613897	98	100	100
FANCG	Fanconi anemia, complementation group G, 614082	602956	100	100	100
FANCI	Fanconi anemia, complementation group I, 609053	611360	53	98	92
FANCL	Fanconi anemia, complementation group L, 614083	608111	52	100	95
FANCM	No OMIM phenotype	609644	55	100	96
FARS2	Combined oxidative phosphorylation deficiency 14, 614946	611592	101	100	100
FAS	{Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic Autoimmune lymphoproliferative syndrome, type IA, 601859	134637	92	100	98
FASLG	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980	134638	87	100	97
FAT4	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006	612411	78	100	98
FBLN1	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180	135820	93	99	96
FBLN5	Cutis laxa, autosomal recessive, type IA, 219100 Cutis laxa, autosomal dominant 2, 614434 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895	604580	75	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FBN1	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370 Geleophysic dysplasia 2, 614185	134797	86	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	90	100	98
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700	611570	99	100	100
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	605654	64	100	100
FBXO38	Neuronopathy, distal hereditary motor, type IID, 615575	608533	56	97	86
FBXO7	Parkinson disease 15, autosomal recessive, 260300	605648	100	100	95
FCGR3A	Immunodeficiency 20, 615707	146740	70	100	96
FCGR3B	Neutropenia, alloimmune neonatal	610665	55	98	93
FCN3	Immunodeficiency due to ficolin 3 deficiency, 613860	604973	82	100	100
FECH	Protoporphyria, erythropoietic, autosomal recessive, 177000	612386	108	100	100
FERMT1	Kindler syndrome, 173650	607900	60	99	95
FERMT3	Leukocyte adhesion deficiency, type III, 612840	607901	76	100	100
FGA	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400	134820	84	100	100
FGB	Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400 Hypofibrinogenemia, congenital, 202400	134830	83	100	100
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	43	99	87
FGD4	Charcot-Marie-Tooth disease, type 4H, 609311	611104	63	100	94
FGF10	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730	602115	87	100	100
FGF14	Spinocerebellar ataxia 27, 609307	601515	96	100	100
FGF16	Metacarpal 4-5 fusion, 309630	300827	30	97	82
FGF17	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270	603725	66	100	100
FGF23	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900	605380	56	100	100
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706	164950	60	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702	600483	78	95	92
FGF9	?Multiple synostoses syndrome 3, 612961	600921	106	100	100
FGFR1	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465	136350	99	100	100
FGFR2	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Scaphocephaly and Axenfeld-Rieger anomaly LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592	176943	80	100	98
FGFR3	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500 Cervical cancer, somatic, 603956 LADD syndrome, 149730 CATSHL syndrome, 610474 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Spermatocytic seminoma, somatic, 273300 SADDAN, 616482	134934	73	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FGG	Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400 Hypodysfibrinogenemia, 616004	134850	86	100	100
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	84	97	91
FHL1	Scapuloperoneal myopathy, X-linked dominant, 300695 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696	300163	62	97	92
FIG4	Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691	609390	86	100	99
FIGLA	Premature ovarian failure 6, 612310	608697	68	97	90
FKBP10	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450	607063	92	100	96
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557	614505	66	100	100
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	72	100	100
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	75	100	100
FLCN	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500	607273	100	100	100
FLG	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803	135940	356	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FLNA	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048	300017	55	100	99
FLNB	Spondylocarpotarsal synostosis syndrome, 272460 Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310	603381	91	100	99
FLNC	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065	102565	90	100	99
FLRT3	Hypogonadotropic hypogonadism 21 with anosmia, 615271	604808	64	100	100
FLT3	Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626 Leukemia, acute lymphoblastic, somatic, 613065	136351	71	99	96
FLT4	Lymphedema, hereditary, IA, 153100 Hemangioma, capillary infantile, somatic, 602089	136352	91	100	99
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	82	100	97
FLVCR2	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790	610865	85	100	100
FMO3	Trimethylaminuria, 602079	136132	93	100	100
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	44	100	89
FN1	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101	135600	82	100	100
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	94	100	100
FOXC1	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631	601090	46	99	75
FOXC2	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400	602402	68	100	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FOXE1	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534	602617	71	98	82
FOXE3	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256	601094	40	73	58
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380	601089	73	100	92
FOXG1	Rett syndrome, congenital variant, 613454	164874	80	83	77
FOXI1	Enlarged vestibular aqueduct, 600791	601093	84	100	100
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996	605597	70	97	90
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705	600838	85	100	99
FOXO1	Rhabdomyosarcoma, alveolar, 268220	136533	89	100	95
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	80	100	98
FOXP2	Speech-language disorder-1, 602081	605317	76	100	100
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100	300292	41	100	94
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	613622	74	100	100
FRAS1	Fraser syndrome, 219000	607830	91	100	100
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	88	100	99
FREM2	Fraser syndrome, 219000	608945	92	100	100
FRMD7	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700	300628	42	100	96
FSCN2	Retinitis pigmentosa 30, 607921	607643	82	100	100
FSHB	Hypogonadotropic hypogonadism 24 without anosmia, 229070	136530	52	100	100
FSHR	Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115	136435	60	100	100
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	57	95	90
FTL	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604	134790	62	94	76
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	70	100	100
FTSJ1	Mental retardation, X-linked 9, 309549	300499	59	100	100
FUCA1	Fucosidosis, 230000	612280	96	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782	137070	79	100	99
FUT6	Fucosyltransferase 6 deficiency, 613852	136836	135	100	100
FUZ	Neural tube defects, 182940	610622	80	100	100
FXN	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300	606829	38	96	72
FXYD2	Hypomagnesemia 2, renal, 154020	601814	49	100	91
FYCO1	Cataract 18, autosomal recessive, 610019	607182	85	100	100
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	93	100	100
FZD6	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157	603409	97	100	100
G6PC	Glycogen storage disease Ia, 232200	613742	115	100	100
G6PC3	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541	611045	65	100	98
G6PD	Hemolytic anemia due to G6PD deficiency, 300908 Favism, 134700 {Resistance to malaria due to G6PD deficiency}, 611162	305900	55	100	98
GAA	Glycogen storage disease II, 232300	606800	81	100	100
GABRA1	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	95	100	100
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269	137192	94	96	94
GABRG2	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681 Febrile seizures, familial, 8, 611277	137164	107	99	92
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	78	100	100
GALC	Krabbe disease, 245200	606890	64	100	98
GALE	Galactose epimerase deficiency, 230350	606953	84	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	76	100	100
GALNS	Mucopolysaccharidosis IVA, 253000	612222	73	98	92
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 211900	601756	72	100	100
GALT	Galactosemia, 230400	606999	120	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	59	99	89
GAN	Giant axonal neuropathy-1, 256850	605379	87	100	100
GARS	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794	600287	75	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GATA1	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835	305371	51	100	100
GATA2	Immunodeficiency 21, 614172 Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 {Leukemia, acute myeloid, susceptibility to}, 601626	137295	69	100	100
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255	131320	86	100	100
GATA4	Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430 ?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500	600576	60	77	62
GATA6	Atrioventricular septal defect 5, 614474 Atrial septal defect 9, 614475 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500	601656	55	85	72
GATAD1	?Cardiomyopathy, dilated, 2B, 614672	614518	91	100	94
GATAD2B	Mental retardation, autosomal dominant 18, 615074	614998	40	98	82
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	93	100	100
GBA	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013 {Parkinson disease, late-onset, susceptibility to}, 168600 {Lewy body dementia, susceptibility to}, 127750	606463	110	100	100
GBA2	Spastic paraplegia 46, autosomal recessive, 614409	609471	62	99	93
GBE1	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	607839	79	100	100
GCDH	Glutaricaciduria, type I, 231670	608801	81	100	99
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	63	100	87
GCK	MODY, type II, 125851 Diabetes mellitus, noninsulin-dependent, late onset, 125853 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176	138079	84	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446	606857	78	100	100
GCM2	Hypoparathyroidism, familial isolated, 146200	603716	77	100	99
GCNT2	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700 Adult i phenotype without cataract, 110800	600429	105	100	100
GCSH	Glycine encephalopathy, 605899	238330	14	61	18
GDAP1	Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340	606598	96	100	100
GDF1	Double-outlet right ventricle, 217095 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854 Right atrial isomerism, 208530	602880	27	70	49
GDF2	Telangiectasia, hereditary hemorrhagic, type 5, 615506	605120	70	100	99
GDF3	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	101	100	100
GDF5	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Brachydactyly, type A2, 112600 Symphalangism, proximal, 1B, 615298 Multiple synostoses syndrome 2, 610017 {Osteoarthritis-5}, 612400 Brachydactyly, type A1, C, 615072	601146	104	100	100
GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703 Leber congenital amaurosis 17, 615360	601147	86	100	100
GDI1	Mental retardation, X-linked 41, 300849	300104	64	100	98
GDNF	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711	600837	144	100	100
GFAP	Alexander disease, 203450	137780	62	100	100
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	58	100	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GFI1	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847	600871	65	100	97
GFI1B	Bleeding disorder, platelet-type, 17, 187900	604383	104	100	100
GFM1	Combined oxidative phosphorylation deficiency 1, 609060	606639	75	100	97
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, 610542	138292	67	100	99
GGCX	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842	137167	86	100	100
GH1	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650	139250	82	100	100
GHR	Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890 Increased responsiveness to growth hormone Growth hormone insensitivity, partial, 604271	600946	77	100	99
GHRHR	Growth hormone deficiency, isolated, type IB, 612781	139191	75	100	98
GHSR	Growth hormone deficiency, isolated partial, 615925	601898	80	100	100
GIF	Intrinsic factor deficiency, 261000	609342	103	100	100
GIGYF2	{Parkinson disease 11}, 607688	612003	65	100	97
GIPC3	Deafness, autosomal recessive 15, 601869	608792	70	94	88
GJA1	Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Hypoplastic left heart syndrome 1, 241550 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, autosomal recessive, 257850 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Palmoplantar keratoderma with congenital alopecia, 104100	121014	66	100	100
GJA3	Cataract 14, multiple types, 601885	121015	95	100	97
GJA5	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770	121013	121	100	100
GJA8	Cataract 1, multiple types, 116200	600897	97	100	100
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800	304040	54	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GJB2	Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540 Bart-Pumphrey syndrome, 149200	121011	122	100	100
GJB3	Erythrokeratodermia variabilis et progressiva, 133200 Deafness, autosomal dominant 2B, 612644 Deafness, autosomal recessive Deafness, autosomal dominant, with peripheral neuropathy Deafness, digenic, GJB2/GJB3, 220290	603324	114	100	100
GJB4	Erythrokeratodermia variabilis with erythema gyratum repens, 133200	605425	120	100	100
GJB6	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500	604418	90	100	100
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480	608803	40	83	58
GK	Glycerol kinase deficiency, 307030	300474	33	99	74
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	64	100	98
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	77	100	97
GLDC	Glycine encephalopathy, 605899	238300	36	81	62
GLE1	Lethal congenital contracture syndrome 1, 253310 Arthrogryposis, lethal, with anterior horn cell disease, 611890	603371	79	100	100
GLI2	Holoprosencephaly-9, 610829 Culler-Jones syndrome, 615849	165230	82	99	97
GLI3	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200 {Hypothalamic hamartomas, somatic}, 241800	165240	88	100	100
GLIS2	Nephronophthisis 7, 611498	608539	74	100	96
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199	610192	84	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GLMN	Glomuvenous malformations, 138000	601749	55	100	96
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400	138491	73	100	99
GLRB	Hyperekplexia 2, autosomal recessive, 614619	138492	65	100	99
GLRX5	?Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950	609588	57	100	86
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	25	80	50
GLUL	Glutamine deficiency, congenital, 610015	138290	79	100	100
GLYCTK	D-glyceric aciduria, 220120	610516	92	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	94	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	57	100	95
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351	615320	100	100	100
GMPS	No OMIM phenotype	600358	70	93	86
GNA11	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361	139313	64	100	98
GNAI2	Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605	139360	79	100	100
GNAI3	Auriculocondylar syndrome 1, 602483	139370	65	100	90
GNAL	Dystonia 25, 615073	139312	49	96	83
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	66	100	99
GNAQ	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000	600998	35	69	47
GNAS	Pseudohypoparathyroidism Ia, 103580 McCune-Albright syndrome, somatic, mosaic 174800 Pseudohypoparathyroidism Ic, 612462 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 Acromegaly, somatic, 102200 Pseudopseudohypoparathyroidism, 612463 ACTH-independent macronodular adrenal hyperplasia, 219080	139320	91	100	97
GNAT1	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389	139330	96	100	100
GNAT2	Achromatopsia-4, 613856	139340	100	100	98
GNB4	Charcot-Marie-Tooth disease, dominant intermediate F, 615185	610863	53	100	90
GNE	Sialuria, 269921 Nonaka myopathy, 605820	603824	86	100	100
GNMT	Glycine N-methyltransferase deficiency, 606664	606628	92	100	98
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	92	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GNPTAB	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500	607840	96	100	97
GNPTG	Mucolipidosis III gamma, 252605	607838	87	95	88
GNRH1	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841	152760	49	100	100
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia, 146110	138850	98	100	100
GNS	Mucopolysaccharidosis type IIID, 252940	607664	84	100	93
GOLGA5	No OMIM phenotype	606918	92	100	100
GORAB	Geroderma osteodysplasticum, 231070	607983	76	100	100
GOSR2	Epilepsy, progressive myoclonic 6, 614018	604027	76	100	100
GOT1	Aspartate aminotransferase, serum level of, QTL1, 614419	138180	90	100	100
GP1BA	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820	606672	82	98	94
GP1BB	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200	138720	31	84	67
GP6	Bleeding disorder, platelet-type, 11, 614201	605546	65	100	100
GP9	Bernard-Soulier syndrome, type C, 231200	173515	75	100	97
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	38	98	92
GPC6	Omodysplasia 1, 258315	604404	89	100	99
GPD1	Hypertriglyceridemia, transient infantile, 614480	138420	78	100	100
GPD1L	Brugada syndrome 2, 611777	611778	88	100	94
GPHN	Molybdenum cofactor deficiency C, 615501	603930	80	100	98
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470	172400	87	100	100
GPR143	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814	300808	36	99	75
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565	614515	78	100	100
GPSM2	Chudley-McCullough syndrome, 604213	609245	76	100	97
GRHL2	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029	608576	75	100	99
GRHL3	Van der Woude syndrome 2, 606713	608317	60	100	96
GRHPR	Hyperoxaluria, primary, type II, 260000	604296	108	100	100
GRIA3	Mental retardation, X-linked 94, 300699	305915	54	100	98
GRIK2	Mental retardation, autosomal recessive, 6, 611092	138244	88	100	99
GRIN1	Mental retardation, autosomal dominant 8, 614254	138249	96	100	99
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	80	100	100
GRIN2B	Mental retardation, autosomal dominant 6, 613970 Epileptic encephalopathy, early infantile, 27, 616139	138252	95	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GRIP1	Fraser syndrome, 219000	604597	82	100	100
GRK1	Oguchi disease-2, 613411	180381	95	100	100
GRM1	Spinocerebellar ataxia, autosomal recessive 13, 614831	604473	89	100	100
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270	604096	73	92	81
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706	138945	95	100	100
GRXCR1	Deafness, autosomal recessive 25, 613285	613283	93	100	100
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471	138890	87	100	98
GSN	Amyloidosis, Finnish type, 105120	137350	82	99	94
GSS	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130	601002	80	100	99
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	112	100	100
GUCA1A	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093	600364	135	100	100
GUCA1B	Retinitis pigmentosa 48, 613827	602275	89	100	100
GUCY1A3	Moyamoya 6 with achalasia, 615750	139396	51	97	89
GUCY2C	Diarrhea 6, 614616 Meconium ileus, 614665	601330	81	100	100
GUCY2D	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777	600179	64	100	93
GUSB	Mucopolysaccharidosis VII, 253220	611499	95	100	99
GYG1	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199	603942	69	96	92
GYS1	Glycogen storage disease 0, muscle, 611556	138570	77	100	100
GYS2	Glycogen storage disease 0, liver, 240600	138571	75	100	95
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data		
H6PD	Cortisone reductase deficiency 1, 604931	138090	100	99	99
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	85	100	100
HADHA	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016	600890	83	100	98
HADHB	Trifunctional protein deficiency, 609015	143450	65	100	95
HAMP	Hemochromatosis, type 2B, 613313	606464	105	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HARS	Usher syndrome type 3B, 614504 Charcot-Marie-Tooth disease, axonal, type 2W, 616625	142810	86	100	99
HARS2	?Perrault syndrome 2, 614926	600783	107	100	100
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738	605998	100	100	100
HBA1	Thalassemias, alpha-, 604131 Methemoglobinemias, alpha- Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978	141800	46	100	95
HBA2	Thalassemia, alpha-, 604131 Heinz body anemia, 140700 Erythrocytosis Hypochromic microcytic anemia Hemoglobin H disease, nondeletional, 613978	141850	45	87	77
HBB	Sickle cell anemia, 603903 Thalassemias, beta-, 613985 Erythremias, beta- Methemoglobinemias, beta- Heinz body anemias, beta-, 140700 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141749 Delta-beta thalassemia, 141749 {Malaria, resistance to}, 611162	141900	165	100	100
HBD	Thalassemia, delta- Thalassemia due to Hb Lepore	142000	154	100	100
HBG1	Fetal hemoglobin quantitative trait locus 1, 141749	142200	57	99	76
HBG2	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977	142250	118	100	100
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	53	100	99
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	44	100	94
HCN4	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123	605206	70	100	97
HCRT	?Narcolepsy 1, 161400	602358	80	100	100
HDAC4	No OMIM phenotype	605314	73	100	100
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	300272	53	100	97
HDAC8	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882	300269	41	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	63	99	83
HERC2	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 Mental retardation, autosomal recessive 38, 615516	605837	37	73	63
HES7	Spondylocostal dysostosis 4, autosomal recessive, 613686	608059	32	80	62
HESX1	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230	601802	78	100	100
HEXA	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800	606869	85	100	100
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	88	100	95
HFE	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferrin serum level QTL2], 614193	613609	77	100	100
HFE2	Hemochromatosis, type 2A, 602390	608374	89	100	100
HFM1	Premature ovarian failure 9, 615724	615684	18	77	36
HGD	Alkaptonuria, 203500	607474	84	100	100
HGF	Deafness, autosomal recessive 39, 608265	142409	83	100	99
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	69	94	91
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	610690	40	87	68
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200	601314	39	88	69
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285	142600	91	100	98
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	88	100	100
HMBS	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000	609806	80	100	100
HMGCL	HMG-CoA lyase deficiency, 246450	613898	98	100	100
HMGCS2	HMG-CoA synthase-2 deficiency, 605911	600234	112	100	100
HMOX1	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	141250	85	100	96
HMX1	Oculoauricular syndrome, 612109	142992	37	77	59

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HNF1A	MODY, type III, 600496 {Diabetes mellitus, noninsulin-dependent, 2}, 125853 {Diabetes mellitus, insulin-dependent}, 222100 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520	142410	82	100	99
HNF1B	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853 {Renal cell carcinoma}, 144700	189907	79	100	98
HNF4A	MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026	600281	80	100	100
HNRNPA1	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426	164017	25	75	51
HOGA1	Hyperoxaluria, primary, type III, 613616	613597	100	100	97
HOXA1	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536	142955	108	100	100
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432	142958	41	100	99
HOXA13	Hand-foot-uterus syndrome, 140000 Guttmacher syndrome, 176305	142959	60	80	72
HOXB1	Facial paresis, hereditary congenital, 3, 614744	142968	75	100	100
HOXC13	Ectodermal dysplasia 9, hair/nail type, 614931	142976	86	100	95
HOXD10	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950	142984	97	100	100
HOXD13	Synpolydactyly, type II, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 Synpolydactyly with foot anomalies, 186000 Syndactyly, type V, 186300 ?Brachydactyly-syndactyly syndrome, 610713	142989	77	100	96
HPD	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350	609695	102	100	100
HPGD	Cranioosteoarthropathy, 259100 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Digital clubbing, isolated congenital, 119900	601688	62	100	100
HPRT1	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323	308000	36	100	85
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	85	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HPS3	Hermansky-Pudlak syndrome 3, 614072	606118	89	100	100
HPS4	Hermansky-Pudlak syndrome 4, 614073	606682	82	100	100
HPS5	Hermansky-Pudlak syndrome 5, 614074	607521	82	100	99
HPS6	Hermansky-Pudlak syndrome 6, 614075	607522	65	99	89
HPSE2	Urofacial syndrome 1, 236730	613469	79	100	100
HR	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550	602302	61	98	95
HRAS	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Spitz nevus or nevus spilus, somatic}, 137550	190020	128	100	100
HRG	Thrombophilia due to HRG deficiency, 613116 Thrombophilia due to elevated HRG, 613116	142640	114	94	94
HSD11B1	Cortisone reductase deficiency 2, 614662	600713	112	100	100
HSD11B2	Apparent mineralocorticoid excess, 218030	614232	75	89	83
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220	300256	63	100	100
HSD17B3	Pseudohermaphroditism, male, with gynecomastia, 264300	605573	73	100	100
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	63	99	96
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810	613890	136	100	100
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765	607764	74	100	100
HSF4	Cataract 5, multiple types, 116800	602438	66	99	95
HSPB1	Neuropathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595	602195	82	92	78
HSPB3	?Neuronopathy, distal hereditary motor, type IIC, 613376	604624	125	100	100
HSPB8	Neuropathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673	608014	118	100	100
HSPD1	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233	118190	27	84	58
HSPG2	Schwartz-Jampel syndrome, type 1, 255800 Dyssegmental dysplasia, Silverman-Handmaker type, 224410	142461	74	100	98
HTR1A	Periodic fever, menstrual cycle dependent, 614674	109760	87	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
HTRA1	{Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149 CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779	602194	76	83	78
HTRA2	{Parkinson disease 13}, 610297	606441	86	100	100
HTT	Huntington disease, 143100	613004	95	99	98
HUWE1	Mental retardation, X-linked syndromic, Turner type, 300706	300697	43	99	89
HYAL1	?Mucopolysaccharidosis type IX, 601492	607071	53	100	100
HYDIN	Ciliary dyskinesia, primary, 5, 608647	610812	147	100	100
HYLS1	Hydrolethalus syndrome, 236680	610693	66	100	100
ICK	Endocrine-cerebroosteodysplasia, 612651	612325	76	100	100
ICOS	Immunodeficiency, common variable, 1, 607594	604558	77	100	100
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	79	100	98
IDH3B	Retinitis pigmentosa 46, 612572	604526	80	100	100
IDS	Mucopolysaccharidosis II, 309900	300823	62	100	100
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015	252800	67	92	82
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	60	78	78
IFITM5	Osteogenesis imperfecta, type V, 610967	614757	52	100	100
IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR, 209950 {H. pylori infection, susceptibility to}, 600263 {Tuberculosis, susceptibility to}, 607948 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {Tuberculosis infection, protection against}, 607948 {Hepatitis B virus infection, susceptibility to}, 610424	107470	65	100	99
IFT122	Cranioectodermal dysplasia 1, 218330	606045	97	100	99
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	82	100	99
IFT172	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Retinitis pigmentosa 71, 616394	607386	77	100	99
IFT43	Cranioectodermal dysplasia 3, 614099	614068	45	100	99
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	611177	54	99	92
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	56	100	100
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	84	100	100
IGF1R	Insulin-like growth factor I, resistance to, 270450	147370	86	100	100
IGF2R	Hepatocellular carcinoma, somatic, 114550	147280	87	98	98
IGFALS	Acid-labile subunit, deficiency of, 615961	601489	59	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224	602867	51	93	82
IGHMBP2	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155	600502	87	100	99
IGLL1	Agammaglobulinemia 2, 613500	146770	36	100	96
IGSF1	Hypothyroidism, central, and testicular enlargement, 300888	300137	55	100	98
IHH	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500	600726	79	100	100
IKBKAP	Dysautonomia, familial, 223900	603722	84	100	99
IKBKB	Immunodeficiency 15, 615592	603258	75	100	99
IKBKG	Incontinentia pigmenti, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 Immunodeficiency 33, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	25	75	49
IKZF1	Leukemia, acute lymphoblastic	603023	91	100	100
IKZF5	No OMIM phenotype	606238	74	100	100
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148	146933	99	100	100
IL10RB	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, autosomal recessive, 612567	123889	106	100	95
IL11RA	Craniosynostosis and dental anomalies, 614188	600939	80	100	100
IL17F	?Candidiasis, familial, 6, autosomal dominant, 613956	606496	95	100	100
IL17RA	?Candidiasis, familial, 5, autosomal recessive, 613953	605461	90	100	96
IL17RD	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267	606807	56	98	90
IL1RAPL1	Mental retardation, X-linked 21/34, 300143	300206	50	100	100
IL1RN	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852	147679	89	100	99
IL21R	[IgE, elevated level of], 147050 Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207	605383	83	100	100
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942	147730	114	100	100
IL2RG	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863	308380	50	100	90
IL31RA	Amyloidosis, primary localized cutaneous, 2, 613955	609510	95	100	100
IL36RN	Psoriasis 14, pustular, 614204	605507	70	100	100
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971	146661	69	100	100
ILDR1	Deafness, autosomal recessive 42, 609646	609739	63	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type, 614078	614010	98	100	98
IMPDH1	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837	146690	72	97	92
IMPG2	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152	607056	89	100	100
INF2	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455	610982	69	92	87
ING1	Squamous cell carcinoma, head and neck, somatic, 275355	601566	62	100	97
INPP5E	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300	613037	71	98	92
INPPL1	Opsismodysplasia, 258480	600829	55	98	89
INS	Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, insulin-dependent, 2, 125852	176730	74	100	100
INSL3	Cryptorchidism, 219050	146738	39	100	81
INSR	Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968	147670	92	98	97
INVS	Nephronophthisis 2, infantile, 602088	243305	82	100	99
IQCB1	Senior-Loken syndrome 5, 609254	609237	65	99	89
IQSEC2	Mental retardation, X-linked 1, 309530	300522	38	94	84
IRAK4	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799	606883	68	100	99
IRF1	Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980	147575	115	100	100
IRF4	[Skin/hair/eye pigmentation, variation in, 8], 611724	601900	116	100	100
IRF6	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 {Orofacial cleft 6}, 608864	607199	79	100	99
IRF8	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894	601565	79	100	100
IRGM	{Mycobacterium tuberculosis, protection against}, 607948 Inflammatory bowel disease 19, 612278	608212	60	100	100
IRX5	Hamamy syndrome, 611174	606195	69	100	92
ISCU	Myopathy with lactic acidosis, hereditary, 255125	611911	68	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	50	97	88
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385	606409	81	95	92
ITGA2B	Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related Bleeding disorder, platelet-type, 16, autosomal dominant, 187800	607759	68	100	100
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748	605025	85	100	97
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730	147556	86	100	100
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	600536	78	97	96
ITGA8	Renal hypodysplasia/aplasia 1, 191830	604063	44	98	86
ITGB2	Leukocyte adhesion deficiency, 116920	600065	114	100	100
ITGB3	Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446 Purpura, posttransfusion Bleeding disorder, platelet-type, 16, autosomal dominant, 187800	173470	79	100	97
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa of hands and feet, 131800	147557	85	100	97
ITK	Lymphoproliferative syndrome 1, 613011	186973	78	100	99
ITM2B	Dementia, familial British, 176500 Dementia, familial Danish, 117300 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079	603904	70	100	100
ITPR1	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	96	100	100
IVD	Isovaleric acidemia, 243500	607036	81	100	99
IYD	Thyroid dyshormonogenesis 4, 274800	612025	91	100	100
JAG1	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon	601920	89	99	96
JAK2	Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 {Budd-Chiari syndrome, somatic}, 600800	147796	65	96	95
JAK3	SCID, autosomal recessive, T-negative/B-positive type, 600802	600173	71	100	98
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	102	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
JPH2	Cardiomyopathy, hypertrophic, 17, 613873	605267	78	100	94
JPH3	Huntington disease-like 2, 606438	605268	81	100	100
JUP	Naxos disease, 601214 Arrhythmogenic right ventricular dysplasia 12, 611528	173325	70	100	100
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900	607704	81	100	100
KANSL1	Koolen-De Vries syndrome, 610443	612452	65	97	86
KARS	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916	601421	100	100	100
KAT6B	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170	605880	94	100	99
KBTBD13	Nemaline myopathy 6, autosomal dominant, 609273	613727	75	100	92
KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	81	100	100
KCNA5	Atrial fibrillation, familial, 7, 612240	176267	92	100	100
KCNC3	Spinocerebellar ataxia 13, 605259	176264	56	80	66
KCND3	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399	605411	104	100	99
KCNE1	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695	176261	163	100	100
KCNE2	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493	603796	103	100	100
KCNE3	Brugada syndrome 6, 613119	604433	88	100	100
KCNH2	Long QT syndrome 2, 613688 {Long QT syndrome 2, acquired, susceptibility to}, 613688 Short QT syndrome 1, 609620	152427	74	98	94
KCNJ1	Bartter syndrome, type 2, 241200	600359	113	100	100
KCNJ10	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791	602208	122	100	100
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582 Maturity-onset diabetes of the young, type 13, 616329	600937	104	100	100
KCNJ13	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186	603208	88	100	100
KCNJ2	Andersen syndrome, 170390 Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980	600681	62	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KCNJ5	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677	600734	102	100	100
KCNK3	Pulmonary hypertension, primary, 4, 615344	603220	84	100	100
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292	605874	88	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	73	100	97
KCNQ1	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Atrial fibrillation, familial, 3, 607554 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500	607542	75	93	90
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data		
KCNQ2	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720	602235	70	100	100
KCNQ3	Seizures, benign neonatal, type 2, 121201	602232	85	100	96
KCNQ4	Deafness, autosomal dominant 2A, 600101	603537	98	95	94
KCNT1	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005	608167	86	100	99
KCNV2	Retinal cone dystrophy 3B, 610356	607604	74	100	100
KCTD1	Scalp-ear-nipple syndrome, 181270	613420	61	90	78
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	90	100	100
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534	314690	52	100	96
KDM6A	Kabuki syndrome 2, 300867	300128	36	95	84
KDR	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089	191306	92	100	100
KERA	Cornea plana congenita, recessive, 217300	603288	91	100	100
KHDC3L	Hydatidiform mole, recurrent, 2, 614293	611687	113	100	100
KIAA0196	Spastic paraplegia 8, autosomal dominant, 603563 Ritscher-Schinzel syndrome 1, 220210	610657	96	100	99
KIAA2022	Mental retardation, X-linked 98, 300912	300524	44	100	97
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	53	99	95
KIF1A	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255	601255	75	100	98
KIF1B	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700	605995	88	100	99
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	No coverage data		
KIF1C	Spastic ataxia 2, autosomal recessive, 611302	603060	55	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700	608283	79	100	96
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546	603213	98	100	100
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	36	87	63
KIF5A	Spastic paraplegia 10, autosomal dominant, 604187	602821	81	100	100
KIF7	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131	611254	64	98	93
KIRREL3	Mental retardation, autosomal dominant 4, 612581	607761	77	100	99
KISS1	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842	603286	46	100	97
KISS1R	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400	604161	68	100	100
KIT	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626 Germ cell tumors, 273300	164920	87	100	100
KITLG	[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 Hyperpigmentation with or without hypopigmentation, 145250 Deafness, congenital, unilateral or asymmetric, 616697	184745	55	100	95
KL	{Coronary artery disease, susceptibility to} Tumoral calcinosis, hyperphosphatemic, 211900	604824	89	98	97
KLF1	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Dyserythropoietic anemia, congenital, type IV, 613673	600599	65	100	97
KLF11	Maturity-onset diabetes of the young, type VII, 610508	603301	96	100	100
KLF6	Prostate cancer, somatic, 176807 Gastric cancer, somatic, 613659	602053	93	100	100
KLHDC8B	{Hodgkin lymphoma, susceptibility to}, 236000	613169	80	100	99
KLHL10	Spermatogenic failure 11, 615081	608778	65	100	100
KLHL3	Pseudohypoaldosteronism, type IID, 614495	605775	96	100	100
KLHL40	Nemaline myopathy 8, autosomal recessive, 615348	615340	61	100	99
KLHL41	Nemaline myopathy 9, 615731	607701	67	100	96
KLHL7	Retinitis pigmentosa 42, 612943	611119	78	100	99
KLK4	Amelogenesis imperfecta, type IIA1, 204700	603767	116	100	100
KLKB1	Fletcher factor (prekallikrein) deficiency, 612423	229000	81	100	98
KLLN	Cowden syndrome 4, 615107	612105	74	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KMT2A	Wiedemann-Steiner syndrome, 605130 Leukemia, myeloid/lymphoid or mixed-lineage	159555	55	99	95
KMT2D	Kabuki syndrome 1, 147920	602113	80	100	99
KNL1	Microcephaly 4, primary, autosomal recessive, 604321	609173	No coverage data		
KPTN	Mental retardation, autosomal recessive 41, 615637	615620	54	100	95
KRAS	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Breast cancer, somatic, 114480 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 RAS-associated autoimmune leukoproliferative disorder, 614470	190070	64	100	100
KRIT1	Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 Cavernous malformations of CNS and retina, 116860	604214	68	100	99
KRT1	Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Ichthyosis histrix, Curth-Macklin type, 146590 Palmoplantar keratoderma, nonepidermolytic, 600962 Palmoplantar keratoderma, epidermolytic, 144200 Keratosis palmoplantaris striata III, 607654	139350	82	100	99
KRT10	Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Ichthyosis with confetti, 609165	148080	105	100	95
KRT12	Meesmann corneal dystrophy, 122100	601687	67	100	97
KRT13	White sponge nevus 2, 615785	148065	91	100	100
KRT14	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800	148066	54	96	87
KRT16	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000	148067	40	87	77

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KRT17	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500	148069	25	73	52
KRT18	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600	148070	54	91	79
KRT2	Ichthyosis bullosa of Siemens, 146800	600194	92	100	100
KRT3	Meesmann corneal dystrophy, 122100	148043	73	100	100
KRT4	White sponge nevus 1, 193900	123940	105	100	100
KRT5	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermylysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex, recessive 1, 601001	148040	96	100	100
KRT6A	Pachyonychia congenita 3, 615726	148041	66	92	85
KRT6B	Pachyonychia congenita 4, 615728	148042	68	93	87
KRT6C	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735	612315	55	86	77
KRT74	Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981 ?Ectodermal dysplasia 7, hair/nail type, 614929	608248	87	100	100
KRT8	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600	148060	19	70	28
KRT81	Monilethrix, 158000	602153	88	100	99
KRT83	?Monilethrix, 158000	602765	85	99	93
KRT85	Ectodermal dysplasia 4, hair/nail type, 602032	602767	105	100	100
KRT86	Monilethrix, 158000	601928	89	100	98
KRT9	Palmoplantar keratoderma, epidermolytic, 144200	607606	63	100	94
L1CAM	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100	308840	52	100	100
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	64	100	92
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855	156225	75	100	99
LAMA3	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, generalized atrophic benign, 226650 Laryngoonychocutaneous syndrome, 245660	600805	87	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
LAMA4	Cardiomyopathy, dilated, 1JJ, 615235	600133	83	100	99
LAMB1	Lissencephaly 5, 615191	150240	91	100	100
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049	150325	95	100	100
LAMB3	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Amelogenesis imperfecta, type IA, 104530	150310	82	100	100
LAMC2	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650	150292	93	100	99
LAMC3	Cortical malformations, occipital, 614115	604349	88	100	98
LAMP2	Danon disease, 300257	309060	65	91	90
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein, 610798	610389	127	100	100
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840	603590	91	100	100
LARP7	Alazami syndrome, 615071	612026	23	63	41
LARS2	Perrault syndrome 4, 615300	604544	50	100	97
LBR	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471	600024	69	100	94
LCA5	Leber congenital amaurosis 5, 604537	611408	70	100	97
LCAT	Norum disease, 245900 Fish-eye disease, 136120	606967	76	99	92
LCT	Lactase deficiency, congenital, 223000	603202	81	100	100
LDB3	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493	605906	94	100	96
LDHA	Glycogen storage disease XI, 612933	150000	45	90	67
LDHB	[Lactate dehydrogenase-B deficiency], 614128	150100	40	87	68
LDLR	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890	606945	112	100	100
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive, 603813	605747	85	100	93
LEF1	Sebaceous tumors, somatic	153245	83	100	95
LEFTY2	Left-right axis malformations	601877	31	81	65
LEMD3	Osteopoikilosis, 166700 Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950	607844	71	100	95
LEP	Obesity, morbid, due to leptin deficiency, 614962	164160	95	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
LEPR	Obesity, morbid, due to leptin receptor deficiency, 614963	601007	74	100	98
LFNG	?Spondylocostal dysostosis 3, autosomal recessive, 609813	602576	54	81	77
LGI1	Epilepsy, familial temporal lobe, 1, 600512	604619	86	100	100
LHB	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300	152780	22	77	52
LHCGR	Precocious puberty, male, 176410 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Luteinizing hormone resistance, female, 238320 Leydig cell adenoma, somatic, with precocious puberty, 176410	152790	78	100	91
LHFPL5	Deafness, autosomal recessive 67, 610265	609427	123	100	100
LHX3	Pituitary hormone deficiency, combined, 3, 221750	600577	60	100	92
LHX4	Pituitary hormone deficiency, combined, 4, 262700	602146	79	100	100
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462	607031	85	100	97
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	151443	71	99	96
LIG1	DNA ligase I deficiency	126391	70	100	100
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	76	100	100
LIM2	Cataract 19, multiple types, 615277	154045	66	100	100
LINS1	Mental retardation, autosomal recessive 27, 614340	610350	No coverage data		
LIPA	Wolman disease, 278000 Cholesteryl ester storage disease, 278000	613497	73	100	100
LIPC	[High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025	151670	91	100	100
LIPH	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379	607365	75	100	100
LIPN	Ichthyosis, congenital, autosomal recessive 8, 613943	613924	87	100	100
LITAF	Charcot-Marie-Tooth disease, type 1C, 601098	603795	88	100	100
LMAN1	Combined factor V and VIII deficiency, 227300	601567	65	100	92
LMBR1	Acheiropody, 200500 Polydactyly, preaxial type II, 174500 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750	605522	54	100	93
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380	612625	57	97	93
LMF1	Lipase deficiency, combined, 246650	611761	89	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
LMNA	Emery-Dreifuss muscular dystrophy 2, AD, 181350 Cardiomyopathy, dilated, 1A, 115200 Lipodystrophy, familial partial, 2, 151660 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Charcot-Marie-Tooth disease, type 2B1, 605588 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Mandibuloacral dysplasia, 248370 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Heart-hand syndrome, Slovenian type, 610140 Malouf syndrome, 212112	150330	61	99	96
LMNB1	Leukodystrophy, adult-onset, autosomal dominant, 169500	150340	84	100	98
LMX1B	Nail-patella syndrome, 161200	602575	84	100	98
LOR	Vohwinkel syndrome with ichthyosis, 604117	152445	24	92	38
LOXHD1	Deafness, autosomal recessive 77, 613079	613072	100	100	100
LPAR6	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150	609239	47	100	100
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive, 268200	605518	78	99	95
LPIN2	Majeed syndrome, 609628	605519	69	100	100
LPL	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]	609708	96	100	100
LPP	Lipoma Leukemia, acute myeloid, 601626	600700	69	100	100
LRAT	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341	604863	137	100	100
LRBA	Immunodeficiency, common variable, 8, with autoimmunity, 614700	606453	72	100	96
LRIG2	Urofacial syndrome 2, 615112	608869	50	96	89
LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058	615004	67	100	100
LRP2	Donnai-Barrow syndrome, 222448	600073	89	100	100
LRP4	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304	604270	93	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
LRP5	Osteoporosis-pseudoglioma syndrome, 259770 [Bone mineral density variability 1], 601884 Hyperostosis, endosteal, 144750 van Buchem disease, type 2, 607636 Osteosclerosis, 144750 {Osteoporosis}, 166710 Exudative vitreoretinopathy 4, 601813 Osteopetrosis, autosomal dominant 1, 607634	603506	112	99	98
LRPAP1	Myopia 23, autosomal recessive, 615431	104225	57	98	89
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	76	100	98
LRRC6	Ciliary dyskinesia, primary, 19, 614935	614930	79	100	95
LRRC8A	Agammaglobulinemia 5, 613506	608360	132	100	100
LRRK2	{Parkinson disease 8}, 607060	609007	75	100	97
LRSAM1	Charcot-Marie-Toothe disease, axonal, type 2P, 614436	610933	84	100	100
LRTOMT	Deafness, autosomal recessive 63, 611451	612414	98	100	100
LTBP2	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Weill-Marchesani syndrome 3, recessive, 614819	602091	72	100	99
LTBP3	Dental anomalies and short stature, 601216	602090	70	99	96
LTBP4	Cutis laxa, autosomal recessive, type IC, 613177	604710	82	99	98
LYST	Chediak-Higashi syndrome, 214500	606897	77	99	97
LYZ	Amyloidosis, renal, 105200	153450	62	100	100
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	87	100	92
LZTS1	Esophageal squamous cell carcinoma, 133239	606551	76	100	100
MAD1L1	Lymphoma, somatic Prostate cancer, somatic, 176807	602686	77	99	93
MAF	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088	177075	53	81	74
MAFB	Multicentric carpotarsal osteolysis syndrome, 166300	608968	89	100	100
MAGEL2	Schaaf-Yang syndrome, 615547	605283	70	100	97
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	44	100	89
MAK	Retinitis pigmentosa 62, 614181	154235	97	100	95
MAML2	Mucoepidermoid salivary gland carcinoma	607537	71	100	100
MAMLD1	Hypospadias 2, X-linked, 300758	300120	54	100	99
MAN1B1	Mental retardation, autosomal recessive 15, 614202	604346	91	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	86	100	98
MANBA	Mannosidosis, beta, 248510	609489	84	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MAOA	Brunner syndrome, 300615	309850	39	100	86
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	87	100	100
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	91	100	100
MAP3K1	46XY sex reversal 6, 613762	600982	95	98	93
MAP3K8	Lung cancer, somatic, 211980	191195	72	100	100
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600	157140	55	97	91
MARS2	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430	609728	93	100	100
MARVELD2	Deafness, autosomal recessive 49, 610153	610572	91	100	98
MASP1	3MC syndrome 1, 257920	600521	103	100	99
MASP2	MASP2 deficiency, 613791	605102	86	100	98
MASTL	?Thrombocytopenia-2, 188000	608221	68	100	99
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850	610550	89	100	100
MATN3	Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600 Spondyloepimetaphyseal dysplasia, 608728	602109	54	85	84
MATR3	Amyotrophic lateral sclerosis 21, 606070	164015	56	91	80
MBD5	Mental retardation, autosomal dominant 1, 156200	611472	82	100	100
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918	300294	54	100	99
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200	607397	80	100	100
MC4R	Obesity, autosomal dominant, 601665	155541	79	100	100
MCC	Colorectal cancer, somatic, 114500	159350	92	100	100
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	77	100	100
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	62	100	95
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120	608419	68	100	100
MCFD2	Factor V and factor VIII, combined deficiency of, 613625	607788	61	92	82
MCM4	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981	602638	79	100	100
MCM6	Lactase persistence/nonpersistence, 223100	601806	80	100	100
MCOLN1	Mucolipidosis IV, 252650	605248	95	100	99
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200	607117	67	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MECP2	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Mental retardation, X-linked syndromic, Lubs type, 300260 Rett syndrome, atypical, 312750	300005	52	99	81
MED12	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895	300188	47	100	91
MED13L	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789	608771	77	100	99
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	77	100	100
MED23	Mental retardation, autosomal recessive 18, 614249	605042	72	100	97
MED25	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449	610197	77	100	98
MEF2C	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443	600662	66	100	98
MEFV	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610	608107	95	100	97
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399	612453	89	100	99
MEGF8	Carpenter syndrome 2, 614976	604267	79	100	99
MEN1	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic	613733	75	100	100
MEOX1	Klippel-Feil syndrome 2, 214300	600147	47	97	91
MERTK	Retinitis pigmentosa 38, 613862	604705	86	100	100
MESP2	Spondylocostal dysostosis 2, autosomal recessive, 608681	605195	72	92	86
MET	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705	164860	95	100	100
MFN2	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA, 601152	608507	84	100	100
MFRP	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	606227	74	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	75	100	97
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	87	100	100
MGME1	Mitochondrial DNA depletion syndrome 11, 615084	615076	56	99	96
MGP	Keutel syndrome, 245150	154870	71	100	97
MIB1	Left ventricular noncompaction 7, 615092	608677	44	99	88
MICU1	Myopathy with extrapyramidal signs, 615673	605084	37	93	80
MID1	Opitz GBBB syndrome, type I, 300000	300552	57	100	99
MINPP1	Thyroid carcinoma, follicular, 188470	605391	79	99	89
MIP	Cataract 15, multiple types, 615274	154050	54	100	93
MIR17HG	Feingold syndrome 2, 614326	609415	No coverage data		
MIR184	EDICT syndrome, 614303	613146	No coverage data		
MIR96	Deafness, autosomal dominant 50, 613074	611606	No coverage data		
MITF	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456	156845	54	100	98
MKKS	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231	604896	106	100	100
MKL1	Megakaryoblastic leukemia, acute	606078	61	100	97
MKRN3	Precocious puberty, central, 2, 615346	603856	47	100	98
MKS1	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 615990	609883	73	100	98
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	71	100	96
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320	120436	85	100	98
MLH3	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385 {Endometrial cancer, susceptibility to}, 608089	604395	74	100	99
MLLT10	Leukemia, acute myeloid, 601626	602409	77	98	97
MLLT11	Leukemia, acute myelomonocytic, somatic, 607785	604684	85	100	100
MLPH	Griscelli syndrome, type 3, 609227	606526	72	100	98
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	67	95	92
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	105	100	100
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110	607568	80	100	100
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	135	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410	611935	60	100	97
MMP1	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600	120353	85	100	100
MMP13	Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111	600108	62	100	98
MMP2	Multicentric osteolysis, nodulosis, and arthropathy, 259600	120360	94	100	100
MMP20	Amelogenesis imperfecta, type IIA2, 612529	604629	56	100	98
MMP9	Metaphyseal anadysplasia 2, 613073	120361	82	100	100
MN1	Meningioma, 607174	156100	71	100	96
MNX1	Currarino syndrome, 176450	142994	48	73	68
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	67	93	77
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	73	100	100
MOG	?Narcolepsy 7, 614250	159465	54	100	96
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	78	100	100
MPC1	Mitochondrial pyruvate carrier deficiency, 614741	614738	59	100	100
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	66	100	99
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219	603785	76	100	97
MPI	Congenital disorder of glycosylation, type Ib, 602579	154550	96	100	100
MPL	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450	159530	89	100	99
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	77	100	100
MPO	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}	606989	95	100	100
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	137960	72	100	100
MPZ	Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 605253 Charcot-Marie-Tooth disease, type 2J, 607736 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, dominant intermediate D, 607791	159440	96	100	100
MRAP	Glucocorticoid deficiency 2, 607398	609196	98	100	100
MRE11A	Ataxia-telangiectasia-like disorder, 604391	600814	53	95	85
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582	607118	43	79	63
MRPS16	Combined oxidative phosphorylation deficiency 2, 610498	609204	91	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	75	100	99
MS4A1	Immunodeficiency, common variable, 5, 613495	112210	72	100	100
MSH2	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300	609309	71	100	97
MSH3	Endometrial carcinoma, somatic, 608089	600887	74	100	98
MSH6	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300	600678	73	100	100
MSR1	Prostate cancer, hereditary, 176807 Barrett esophagus/esophageal adenocarcinoma, 614266	153622	78	100	99
MSRB3	Deafness, autosomal recessive 74, 613718	613719	72	100	99
MSTN	Muscle hypertrophy, 614160	601788	81	100	100
MSX1	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500	142983	73	100	96
MSX2	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550	123101	73	100	100
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250	156540	72	90	90
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947	611766	67	100	95
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050	607093	94	100	100
MTM1	Myotubular myopathy, X-linked, 310400	300415	42	99	93
MTMR2	Charcot-Marie-Tooth disease, type 4B1, 601382	603557	78	100	100
MTO1	Combined oxidative phosphorylation deficiency 10, 614702	614667	85	94	93
MTPAP	Ataxia, spastic, 4, 613672	613669	74	100	99
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	80	100	99
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	81	100	100
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	78	100	96
MUC1	Medullary cystic kidney disease 1, 174000	158340	37	95	75
MUSK	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 Fetal akinesia deformation sequence, 208150	601296	93	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	81	100	100
MUTYH	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600	604933	82	100	100
MVK	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900	251170	94	100	100
MXI1	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807	600020	62	100	96
MYBPC1	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915	160794	82	100	100
MYBPC3	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396	600958	90	100	97
MYCN	Feingold syndrome, 164280	164840	68	100	91
MYD88	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600	602170	131	100	100
MYF6	Myopathy, centronuclear, 3, 614408	159991	92	100	100
MYH11	Aortic aneurysm, familial thoracic 4, 132900	160745	90	100	100
MYH14	Deafness, autosomal dominant 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369	608568	70	99	95
MYH2	Proximal myopathy and ophthalmoplegia, 605637	160740	68	100	98
MYH3	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110	160720	78	100	99
MYH6	Cardiomyopathy, hypertrophic, 14, 613251 Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 {Sick sinus syndrome 3}, 614090	160710	92	99	98
MYH7	Cardiomyopathy, hypertrophic, 1, 192600 Cardiomyopathy, dilated, 1S, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Liang distal myopathy, 160500 Scapuloperoneal syndrome, myopathic type, 181430 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal recessive, 255160	160760	90	100	100
MYH8	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300	160741	85	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MYH9	May-Hegglin anomaly, 155100 Fechtner syndrome, 153640 Sebastian syndrome, 605249 Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Macrothrombocytopenia and progressive sensorineural deafness, 600208	160775	91	100	100
MYL2	Cardiomyopathy, hypertrophic, 10, 608758	160781	90	100	100
MYL3	Cardiomyopathy, hypertrophic, 8, 608751	160790	87	100	100
MYLK	Aortic aneurysm, familial thoracic 7, 613780	600922	102	100	100
MYLK2	Cardiomyopathy, hypertrophic, 1, digenic, 192600	606566	84	100	100
MYO15A	Deafness, autosomal recessive 3, 600316	602666	83	98	94
MYO1A	No OMIM phenotype	601478	87	100	100
MYO1E	Glomerulosclerosis, focal segmental, 6, 614131	601479	77	100	99
MYO3A	Deafness, autosomal recessive 30, 607101	606808	70	100	98
MYO5A	Griscelli syndrome, type 1, 214450	160777	74	100	98
MYO5B	Microvillus inclusion disease, 251850	606540	91	100	97
MYO6	Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346	600970	59	100	94
MYO7A	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317	276903	100	100	98
MYOC	Glaucoma 1A, primary open angle, 137750	601652	101	100	100
MYOT	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920	604103	75	100	96
MYOZ2	Cardiomyopathy, hypertrophic, 16, 613838	605602	85	100	100
MYPN	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248	608517	101	100	99
NAA10	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800	300013	51	100	100
NAGA	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241	104170	86	100	100
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491	609701	81	100	93
NAGS	N-acetylglutamate synthase deficiency, 237310	608300	62	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266	611549	43	98	87
NANOS1	Spermatogenic failure 12, 615413	608226	44	96	82
NBAS	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483	608025	79	100	98
NBEAL2	Gray platelet syndrome, 139090	614169	87	100	99
NBN	Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065	602667	66	100	97
NCF1	Chronic granulomatous disease due to deficiency of NCF-1, 233700	608512	10	21	19
NCF2	Chronic granulomatous disease due to deficiency of NCF-2, 233710	608515	83	100	100
NCF4	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960	601488	104	100	100
NCOA4	No OMIM phenotype	601984	91	91	89
NCSTN	Acne inversa, familial, 1, 142690	605254	84	100	97
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	94	100	100
NDN	Prader-Willi syndrome, 176270	602117	74	100	97
NDP	Norrie disease, 310600 Exudative vitreoretinopathy 2, X-linked, 305390	300658	68	100	100
NDRG1	Charcot-Marie-Tooth disease, type 4D, 601455	605262	65	100	100
NDUFA1	Mitochondrial complex I deficiency, 252010	300078	85	100	90
NDUFA10	?Leigh syndrome, 256000	603835	94	99	96
NDUFA11	Mitochondrial complex I deficiency, 252010	612638	57	100	96
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000	614530	98	100	100
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	71	100	100
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency, 256000	603834	73	92	81
NDUFAF1	Mitochondrial complex I deficiency, 252010	606934	81	100	100
NDUFAF2	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000	609653	14	65	23
NDUFAF3	Mitochondrial complex I deficiency, 252010	612911	83	100	100
NDUFAF4	Mitochondrial complex I deficiency, 252010	611776	71	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010	612360	67	100	95
NDUFAF6	Leigh syndrome due to mitochondrial complex I deficiency, 256000	612392	55	100	96
NDUFB3	Mitochondrial complex I deficiency, 252010	603839	47	65	48
NDUFS1	Mitochondrial complex I deficiency, 252010	157655	76	100	98
NDUFS2	Mitochondrial complex I deficiency, 252010	602985	89	100	100
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	603846	126	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010	602694	81	100	100
NDUFS6	Mitochondrial complex I deficiency, 252010	603848	65	100	85
NDUFS7	Leigh syndrome, 256000	601825	77	100	100
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	91	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010	161015	92	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010	600532	30	68	36
NEB	Nemaline myopathy 2, autosomal recessive, 256030	161650	137	100	99
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	600644	No coverage data		
NECTIN4	Ectodermal dysplasia-syndactyly syndrome 1, 613573	609607	No coverage data		
NEFL	Charcot-Marie-Tooth disease, type 2E, 607684 Charcot-Marie-Tooth disease, type 1F, 607734	162280	95	100	100
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604588	62	99	92
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	60	95	86
NEUROD1	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394	601724	66	100	100
NEUROG3	Diarrhea 4, malabsorptive, congenital, 610370	604882	100	100	100
NEXN	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876	613121	49	100	95
NF1	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	80	99	98
NF2	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091	607379	80	100	99
NFIX	Sotos syndrome 2, 614753 Marshall-Smith syndrome, 602535	164005	90	100	100
NFKB2	Immunodeficiency, common variable, 10, 615577	164012	86	100	97
NFKBIA	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132	164008	80	100	98
NFU1	Multiple mitochondrial dysfunctions syndrome 1, 605711	608100	48	91	85
NGF	Neuropathy, hereditary sensory and autonomic, type V, 608654	162030	79	100	100
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291	611290	63	100	99
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	86	100	100
NHP2	Dyskeratosis congenita, autosomal recessive 2, 613987	606470	52	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NHS	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200	300457	48	95	92
NIN	Seckel syndrome 7, 614851	608684	89	100	99
NIPA1	Spastic paraplegia 6, autosomal dominant, 600363	608145	116	100	97
NIPAL4	Ichthyosis, congenital, autosomal recessive 6, 612281	609383	85	100	98
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	64	98	93
NKX2-1	{Thyroid cancer, monmedullary, 1}, 188550 Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978	600635	70	100	97
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, 108900 Tetrology of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095	600584	79	100	100
NKX2-6	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095	611770	101	100	100
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	602183	73	100	100
NLGN4X	{Autism susceptibility, X-linked 2}, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 Mental retardation, X-linked, 300495	300427	74	100	100
NLRP12	Familial cold autoinflammatory syndrome 2, 611762	609648	100	100	100
NLRP3	Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115	606416	102	100	100
NLRP7	Hydatidiform mole, recurrent, 1, 231090	609661	108	100	100
NME8	Ciliary dyskinesia, primary, 6, 610852	607421	76	100	99
NMNAT1	Leber congenital amaurosis 9, 608553	608700	119	100	100
NNT	Glucocorticoid deficiency 4, 614736	607878	68	100	97
NOBOX	Premature ovarian failure 5, 611548	610934	59	100	100
NOD2	{Inflammatory bowel disease 1}, 266600 Blau syndrome, 186580 {Psoriatic arthritis, susceptibility to}, 607507 Sarcoidosis, early-onset, 609464	605956	88	100	100
NODAL	Heterotaxy, visceral, 5, 270100	601265	106	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NOG	Symphalangism, proximal, 1A, 185800 Multiple synostoses syndrome 1, 186500 Tarsal-carpal coalition syndrome, 186570 Stapes ankylosis with broad thumb and toes, 184460 Brachydactyly, type B2, 611377	602991	128	100	100
NOL3	Myoclonus, familial cortical, 614937	605235	54	100	99
NOP10	Dyskeratosis congenita, autosomal recessive 1, 224230	606471	65	100	86
NOP56	Spinocerebellar ataxia 36, 614153	614154	68	96	90
NOTCH1	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028	190198	86	100	99
NOTCH2	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500	600275	89	100	100
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 ?Myofibromatosis, infantile 2, 615293 Lateral meningocele syndrome, 130720	600276	66	95	90
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	83	100	96
NPC2	Niemann-pick disease, type C2, 607625	601015	115	100	100
NPHP1	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583	607100	78	100	99
NPHP3	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010	608002	77	100	100
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	87	100	100
NPHS1	Nephrotic syndrome, type 1, 256300	602716	66	100	97
NPHS2	Nephrotic syndrome, type 2, 600995	604766	69	100	96
NPM1	Leukemia, acute myeloid, somatic, 601626	164040	65	100	93
NPPA	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745	108780	108	100	100
NPR2	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255	108961	100	100	100
NR0B1	Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200 46XY sex reversal 2, dosage-sensitive, 300018	300473	55	100	100
NR0B2	Obesity, mild, early-onset, 601665	604630	64	100	100
NR2E3	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131	604485	65	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	123	100	100
NR3C1	Glucocorticoid resistance, 615962	138040	83	100	100
NR3C2	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115	600983	75	99	96
NR4A3	Chondrosarcoma, extraskeletal myxoid, 612237	600542	79	100	94
NR5A1	46XY sex reversal 3, 612965 Premature ovarian failure 7, 612964 Adrenocortical insufficiency Spermatogenic failure 8, 613957	184757	67	100	99
NRAS	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	164790	78	100	100
NRL	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type	162080	54	100	100
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	102	100	99
NSD1	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650	606681	90	100	99
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	57	100	100
NSMF	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838	608137	59	95	95
NSUN2	Mental retardation, autosomal recessive 5, 611091	610916	78	100	95
NT5C2	Spastic paraplegia 45, autosomal recessive, 613162	600417	44	93	75
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency, 266120	606224	33	81	58
NT5E	Calcification of joints and arteries, 211800	129190	107	100	100
NTF4	Glaucoma 1, open angle, 1O, 613100	162662	73	100	91
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	75	100	99
NTRK2	?Obesity, hyperphagia, and developmental delay, 613886	600456	88	100	100
NUBPL	Mitochondrial complex I deficiency, 252010	613621	55	100	92
NUMA1	Leukemia, acute promyelocytic, somatic, 612376	164009	77	100	99
NUP214	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065	114350	99	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
NUP62	Striatonigral degeneration, infantile, 271930	605815	67	100	100
NYX	Night blindness, congenital stationary (complete), 1A, X-linked, 310500	300278	43	100	97
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	72	100	96
OBSL1	3-M syndrome 2, 612921	610991	81	100	98
OCA2	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220	611409	75	100	98
OCLN	Band-like calcification with simplified gyration and polymicrogyria, 251290	602876	79	100	100
OCRL	Lowe syndrome, 309000 Dent disease 2, 300555	300535	45	99	93
OFD1	Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804 ?Retinitis pigmentosa 23, 300424	300170	43	99	88
OGG1	Renal cell carcinoma, clear cell, somatic, 144700	601982	80	100	100
OPA1	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250	605290	69	100	93
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	72	100	90
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	42	100	97
OPLAH	5-oxoprolinase deficiency, 260005	614243	66	100	97
OPN1LW	Colorblindness, protan, 303900 Blue cone monochromacy, 303700	300822	27	59	46
OPN1MW	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700	300821	22	65	50
OPN1SW	Colorblindness, tritan, 190900	613522	89	100	100
OPTN	Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657 Amyotrophic lateral sclerosis 12, 613435	602432	69	100	93
ORAI1	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883	610277	105	98	93
ORC1	Meier-Gorlin syndrome 1, 224690	601902	94	100	100
ORC4	Meier-Gorlin syndrome 2, 613800	603056	49	100	93
ORC6	Meier-Gorlin syndrome 3, 613803	607213	71	100	100
OSMR	Amyloidosis, primary localized cutaneous, 1, 105250	601743	82	100	99
OSTM1	Osteopetrosis, autosomal recessive 5, 259720	607649	59	93	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
OTC	Ornithine transcarbamylase deficiency, 311250	300461	59	100	93
OTOA	Deafness, autosomal recessive 22, 607039	607038	88	100	98
OTOF	Deafness, autosomal recessive 9, 601071 Auditory neuropathy, autosomal recessive, 1, 601071	603681	90	100	99
OTOG	Deafness, autosomal recessive 18B, 614945	604487	82	100	99
OTOGL	Deafness, autosomal recessive 84B, 614944	614925	68	100	96
OTX2	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125	600037	88	100	100
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	601424	63	100	100
P2RX1	Bleeding disorder due to P2RX1 defect, somatic, 609821	600845	73	100	100
P2RX2	Deafness, autosomal dominant 41, 608224	600844	61	100	95
P2RY12	Bleeding disorder, platelet-type, 8, 609821	600515	93	100	100
P3H1	Osteogenesis imperfecta, type VIII, 610915	610339	87	100	100
P3H2	Myopia, high, with cataract and vitreoretinal degeneration, 614292	610341	79	100	100
PABPN1	Oculopharyngeal muscular dystrophy, 164300	602279	60	64	62
PACS1	Mental retardation, autosomal dominant 17, 615009	607492	78	98	95
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	68	100	97
PAH	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600	612349	112	100	100
PAK3	Mental retardation, X-linked 30/47, 300558	300142	41	99	86
PALB2	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348	610355	85	100	100
PANK2	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236	606157	90	100	100
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847	603005	60	100	95
PARK2	Parkinson disease, juvenile, type 2, 600116 Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 {Leprosy, susceptibility to}, 607572	602544	69	100	99
PARK7	Parkinson disease 7, autosomal recessive early-onset, 606324	602533	63	100	99
PAX2	Papillorenal syndrome, 120330 Glomerulosclerosis, focal segmental, 7, 616002	167409	100	100	98
PAX3	Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820 Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220	606597	71	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PAX4	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227	167413	65	100	100
PAX6	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hypoplasia 1, 136520 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700	607108	62	100	99
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	167415	73	100	98
PAX9	Tooth agenesis, selective, 3, 604625	167416	128	99	99
PC	Pyruvate carboxylase deficiency, 266150	608786	84	100	100
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070	126090	87	100	98
PCCA	Propionicacidemia, 606054	232000	55	99	88
PCCB	Propionicacidemia, 606054	232050	87	100	100
PCDH15	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067	605514	89	100	98
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	57	100	97
PCM1	No OMIM phenotype	600299	75	100	99
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	79	100	97
PCSK1	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362	162150	79	100	100
PCSK9	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776	607786	67	100	97
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	40	96	78
PDCD10	Cerebral cavernous malformations 3, 603285	609118	62	100	90
PDE11A	Pigmented nodular adrenocortical disease, primary, 2, 610475	604961	79	100	96
PDE4D	{Stroke, susceptibility to, 1}, 606799 Acrodysostosis 2, with or without hormone resistance, 614613	600129	64	99	95
PDE6A	Retinitis pigmentosa 43, 613810	180071	75	100	99
PDE6B	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801	180072	101	100	100
PDE6C	Cone dystrophy 4, 613093	600827	75	100	99
PDE6G	Retinitis pigmentosa 57, 613582	180073	88	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PDE6H	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024	601190	48	100	100
PDE8B	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161	603390	66	100	98
PDGFB	Meningioma, SIS-related, 607174 Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483	190040	73	100	100
PDGFRA	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685	173490	92	100	100
PDGFRB	Myeloproliferative disorder with eosinophilia, 131440 Basal ganglia calcification, idiopathic, 4, 615007 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592	173410	92	100	99
PDGFRL	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500	604584	100	100	100
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	64	99	92
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	79	100	100
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	83	100	100
PDS5B	No OMIM phenotype	605333	35	90	73
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	65	91	83
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	69	99	94
PDX1	Pancreatic agenesis 1, 260370 MODY, type IV, 606392 {Diabetes mellitus, type II, susceptibility to}, 125853	600733	37	100	76
PDYN	Spinocerebellar ataxia 23, 610245	131340	76	100	100
PDZD7	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	60	99	96
PEPD	Prolidase deficiency, 170100	613230	68	100	94
PER2	Advanced sleep phase syndrome, familial, 1, 604348	603426	78	100	98
PET100	Mitochondrial complex IV deficiency, 220110	614770	35	97	64
PEX1	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Heimler syndrome 1, 234580	602136	67	100	98
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	60	95	89
PEX11B	Peroxisome biogenesis disorder 14B, 614920	603867	92	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	104	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	91	100	100
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887	601791	85	100	100
PEX16	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	85	100	95
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	75	100	100
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	71	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	94	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882	603164	58	100	98
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	93	100	100
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617	601498	71	95	91
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879	601757	68	100	89
PFKM	Glycogen storage disease VII, 232800	610681	84	100	100
PFN1	Amyotrophic lateral sclerosis 18, 614808	176610	127	100	100
PGAM2	Glycogen storage disease X, 261670	612931	79	100	100
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	63	100	92
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	42	96	81
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	43	100	93
PGM1	Congenital disorder of glycosylation, type It, 614921	171900	99	100	100
PHEX	Hypophosphatemic rickets, X-linked dominant, 307800	300550	42	100	96
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	31	98	73
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263	300560	52	99	90
PHGDH	Phosphoglycerate dehydrogenase deficiency, 601815 Neu-Laxova syndrome 1, 256520	606879	86	100	99
PHKA1	Muscle glycogenosis, 300559	311870	37	93	80
PHKA2	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000	300798	45	100	96
PHKB	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750	172490	69	100	98
PHKG2	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency	172471	72	100	100
PHOX2A	Fibrosis of extraocular muscles, congenital, 2, 602078	602753	39	81	57

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PHOX2B	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013 Neuroblastoma with Hirschsprung disease, 613013	603851	67	99	91
PHRF1	No OMIM phenotype	611780	66	100	95
PHYH	Refsum disease, 266500	602026	57	99	85
PICALM	Leukemia, acute myeloid, somatic, 601626	603025	57	100	98
PIEZO1	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843	611184	61	99	94
PIEZO2	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700	613629	40	97	86
PIGA	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868	311770	41	93	83
PIGL	CHIME syndrome, 280000	605947	74	100	94
PIGM	Glycosylphosphatidylinositol deficiency, 610293	610273	67	100	100
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	66	98	93
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	82	100	100
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	102	100	100
PIK3CA	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Nonsmall cell lung cancer, somatic, 211980 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108	171834	71	100	98
PIK3CD	Immunodeficiency 14, 615513	602839	100	100	99
PIK3R1	?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880 Immunodeficiency 36, 616005	171833	81	100	100
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	70	90	89
PIK3R5	Ataxia-oculomotor apraxia 3, 615217	611317	76	100	100
PIKFYVE	Corneal fleck dystrophy, 121850	609414	88	100	98
PINK1	Parkinson disease 6, early onset, 605909	608309	65	94	90
PIP5K1C	Lethal congenital contractural syndrome 3, 611369	606102	81	96	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PITPNM3	Cone-rod dystrophy 5, 600977	608921	75	99	99
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550	602149	81	100	92
PITX2	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Ring dermoid of cornea, 180550 Peters anomaly, 604229	601542	74	98	91
PITX3	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623	602669	60	100	100
PKD1	Polycystic kidney disease, adult type I, 173900	601313	76	98	95
PKD2	Polycystic kidney disease 2, 613095	173910	67	95	89
PKHD1	Polycystic kidney and hepatic disease, 263200	606702	83	100	99
PKLR	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900	609712	85	100	100
PKP1	Ectodermal dysplasia/skin fragility syndrome, 604536	601975	80	100	99
PKP2	Arrhythmogenic right ventricular dysplasia 9, 609040	602861	80	97	92
PLA2G4A	Phospholipase A2, group IV A, deficiency of	600522	80	100	98
PLA2G5	[Fleck retina, familial benign], 228980	601192	105	100	100
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953	603604	88	100	99
PLA2G7	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050	601690	78	100	100
PLAG1	Adenomas, salivary gland pleomorphic, somatic, 181030	603026	81	100	100
PLAU	{Alzheimer disease, late-onset, susceptibility to}, 104300 Quebec platelet disorder, 601709	191840	84	100	100
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	76	100	99
PLCB4	Auriculocondylar syndrome 2, 614669	600810	73	100	97
PLCD1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600	602142	63	100	99
PLCE1	Nephrotic syndrome, type 3, 610725	608414	90	100	99
PLCG2	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878	600220	94	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PLEC	Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex, Ogna type, 131950 Epidermolysis bullosa simplex with pyloric atresia, 612138 Muscular dystrophy, limb-girdle, type 2Q, 613723 ?Epidermolysis bullosa simplex with nail dystrophy, 616487	601282	75	100	97
PLEKHG5	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376	611101	60	100	94
PLEKHM1	Osteopetrosis, autosomal recessive 6, 611497	611466	52	100	94
PLG	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090	173350	80	93	87
PLIN1	Lipodystrophy, familial partial, type 4, 613877	170290	55	100	95
PLN	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874	172405	147	100	100
PLOD1	Ehlers-Danlos syndrome, type VI, 225400	153454	88	100	98
PLOD2	Bruck syndrome 2, 609220	601865	72	99	92
PLOD3	Lysyl hydroxylase 3 deficiency, 612394	603066	69	100	96
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920	300401	71	100	99
PLS3	Bone mineral density QTL18, osteoporosis, 300910	300131	33	97	80
PML	Leukemia, acute promyelocytic, PML/RARA type	102578	75	100	100
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	93	100	100
PMP22	Charcot-Marie-Tooth disease, type 1A, 118220 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, inflammatory demyelinating, 139393	601097	117	100	100
PMS2	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337	600259	86	100	100
PNKP	Microcephaly, seizures, and developmental delay, 613402 Ataxia-oculomotor apraxia 4, 616267	605610	61	100	98
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	85	100	100
PNPLA1	Ichthyosis, congenital, autosomal recessive 10, 615024	612121	87	100	100
PNPLA2	Neutral lipid storage disease with myopathy, 610717	609059	73	100	99
PNPLA6	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400	603197	72	100	98
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	66	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PNPT1	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934	610316	33	87	63
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	90	100	100
POF1B	Premature ovarian failure 2B, 300604	300603	37	95	82
POFUT1	Dowling-Degos disease 2, 615327	607491	57	100	95
POGLUT1	Dowling-Degos disease 4, 615696	615618	43	98	85
POLD1	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381	174761	73	100	100
POLE	{Colorectal cancer, susceptibility to, 12}, 615083 FILS syndrome, 615139	174762	55	99	92
POLG	Progressive external ophthalmoplegia, autosomal recessive 1, 258450 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459	174763	86	100	100
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131	604983	115	100	97
POLH	Xeroderma pigmentosum, variant type, 278750	603968	76	100	99
POLR1C	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494	610060	92	100	100
POLR1D	Treacher Collins syndrome 2, 613717	613715	73	100	100
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	88	100	100
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	82	100	99
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665	176830	70	100	100
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157	606822	82	100	95
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830	614828	85	100	100
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952	613386	61	100	97
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	90	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	64	98	95
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	124015	94	100	100
PORCN	Focal dermal hypoplasia, 305600	300651	59	100	96
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	71	100	97
POU3F4	Deafness, X-linked 2, 304400	300039	49	100	100
POU4F3	Deafness, autosomal dominant 15, 602459	602460	131	100	100
PPARG	Obesity, severe, 601665 [Obesity, resistance to] Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Carotid intimal medial thickness 1, 609338 {Diabetes, type 2}, 125853	601487	98	100	100
PPIB	Osteogenesis imperfecta, type IX, 259440	123841	107	100	100
PPM1D	Breast cancer, 114480	605100	88	100	100
PPM1K	?Maple syrup urine disease, mild variant, 615135	611065	90	100	99
PPOX	Porphyria variegata, 176200	600923	69	100	100
PPP1CB	No OMIM phenotype	600590	38	98	81
PPP1R3A	Insulin resistance, severe, digenic, 604367	600917	88	100	100
PPP2R1B	Lung cancer, 211980	603113	79	100	96
PPP2R2B	Spinocerebellar ataxia 12, 604326	604325	83	100	100
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	81	100	100
PQBP1	Renpenning syndrome, 309500	300463	62	100	100
PRCC	Renal cell carcinoma, papillary, 605074	179755	83	100	96
PRCD	Retinitis pigmentosa 36, 610599	610598	51	100	100
PRDM16	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373	605557	107	100	98
PRDM5	Brittle cornea syndrome 2, 614170	614161	74	100	99
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027 Aplastic anemia, 609135	170280	74	100	100
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250	604283	86	100	98
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437	608500	73	100	97
PRICKLE2	Epilepsy, progressive myoclonic 5, 613832	608501	80	100	100
PRIMPOL	Myopia 22, autosomal dominant, 615420	615421	40	89	71

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PRKAG2	Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740	602743	78	100	98
PRKAR1A	Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489 Adrenocortical tumor, somatic, Acrodysostosis 1, with or without hormone resistance, 101800	188830	96	100	100
PRKCA	Pituitary tumor, invasive	176960	111	100	100
PRKCG	Spinocerebellar ataxia 14, 605361	176980	84	100	99
PRKCSH	Polycystic liver disease, 174050	177060	77	100	96
PRKG1	Aortic aneurysm, familial thoracic 8, 615436	176894	46	97	82
PRKRA	Dystonia 16, 612067	603424	55	100	97
PRLR	Multiple fibroadenomas of the breast, 615554 ?Hyperprolactinemia, 615555	176761	79	100	99
PRNP	Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 Huntington disease-like 1, 603218 {Kuru, susceptibility to}, 245300 Cerebral amyloid angiopathy, PRNP-related, 137440	176640	51	100	97
PROC	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304	612283	76	100	100
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	51	95	83
PROK2	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628	607002	42	99	82
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200	607123	176	100	100
PROM1	Retinitis pigmentosa 41, 612095 Cone-rod dystrophy 12, 612657 Stargardt disease 4, 603786 Macular dystrophy, retinal, 2, 608051	604365	72	100	99
PROP1	Pituitary hormone deficiency, combined, 2, 262600	601538	48	100	81
PROS1	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514	176880	37	92	76
PRPF3	Retinitis pigmentosa 18, 601414	607301	81	100	100
PRPF31	Retinitis pigmentosa 11, 600138	606419	73	100	99
PRPF6	Retinitis pigmentosa 60, 613983	613979	91	100	100
PRPF8	Retinitis pigmentosa 13, 600059	607300	89	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PRPH2	Retinitis pigmentosa 7 and digenic, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choriodal dystrophy, central areolar 2, 613105 Macular dystrophy, vitelliform, 3, 608161	179605	87	100	100
PRPS1	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500	311850	59	100	100
PRRT2	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066	614386	58	100	100
PRRX1	Agnathia-otocephaly complex, 202650	167420	75	100	100
PRSS1	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044	276000	158	100	100
PRSS12	Mental retardation, autosomal recessive 1, 249500	606709	82	100	98
PRSS56	Microphthalmia, isolated 6, 613517	613858	52	100	99
PRX	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 4F, 614895	605725	92	100	98
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722	176801	98	100	100
PSAT1	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038	610936	33	71	49
PSENEN	Acne inversa, familial, 2, 613736	607632	101	100	100
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040	177046	41	97	83
PSMC3IP	Ovarian dysgenesis 3, 614324	608665	54	100	100
PSPH	Phosphoserine phosphatase deficiency, 614023	172480	49	100	98
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416	606347	67	100	98
PTCH1	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828	601309	88	99	96
PTCH2	Medulloblastoma, 155255 Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400	603673	65	100	99
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	46	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PTEN	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355	601728	57	77	73
PTF1A	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935	607194	75	95	84
PTGIS	Hypertension, essential, 145500	601699	89	95	95
PTH	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200	168450	107	100	100
PTH1R	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350	168468	76	100	100
PTHLH	Humoral hypercalcemia of malignancy Brachydactyly, type E2, 613382	168470	76	100	99
PTPN11	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250	176876	68	99	99
PTPN12	Colon cancer, somatic, 114500	600079	69	100	93
PTPN14	Choanal atresia and lymphedema, 613611	603155	88	100	100
PTPRC	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971	151460	68	98	92
PTPRJ	Colon cancer, somatic, 114500	600925	88	98	97
PTPRO	Nephrotic syndrome, type 6, 614196	600579	78	100	99
PTPRQ	Deafness, autosomal recessive 84A, 613391	603317	69	99	95
PTRF	Lipodystrophy, congenital generalized, type 4, 613327	603198	88	100	100
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	58	100	99
PUF60	Verheij syndrome, 615583	604819	71	97	95
PUS1	Mitochondrial myopathy and sideroblastic anemia 1, 600462	608109	71	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438	179035	71	100	96
PYGL	Glycogen storage disease VI, 232700	613741	91	100	97
PYGM	McArdle disease, 232600	608455	98	100	100
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	57	99	92
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	60	100	95
RAB18	Warburg micro syndrome 3, 614222	602207	59	97	91
RAB23	Carpenter syndrome, 201000	606144	85	100	100
RAB27A	Griscelli syndrome, type 2, 607624	603868	83	100	100
RAB28	Cone-rod dystrophy 18, 615374	612994	20	84	39
RAB33B	Smith-McCort dysplasia 2, 615222	605950	75	100	100
RAB39B	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510	300774	48	100	97
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	68	100	98
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	69	100	96
RAB40AL	No OMIM phenotype	300405	50	100	100
RAB7A	Charcot-Marie-Tooth disease, type 2B, 600882	602298	87	100	100
RAC2	Neutrophil immunodeficiency syndrome, 608203	602049	76	100	100
RAD21	Cornelia de Lange syndrome 4, 614701	606462	65	100	93
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	66	97	92
RAD51	{Breast cancer, susceptibility to}, 114480 Mirror movements 2, 614508	179617	58	88	88
RAD51C	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399	602774	84	100	100
RAD54B	Lymphoma, non-Hodgkin, somatic, 605027 Colon cancer, somatic, 114500	604289	76	100	99
RAD54L	{Breast cancer, invasive ductal}, 114480 Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic	603615	84	100	100
RAF1	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554 Cardiomyopathy, dilated, 1NN, 615916	164760	82	100	98
RAG1	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650	179615	83	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RAG2	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 Combined cellular and humoral immune defects with granulomas, 233650	179616	103	100	100
RAI1	Smith-Magenis syndrome, 182290	607642	91	100	100
RAP1GDS1	Lymphocytic leukemia, acute T-cell	179502	58	98	93
RAPSN	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 Fetal akinesia deformation sequence, 208150	601592	100	100	100
RARB	Microphthalmia, syndromic 12, 615524	180220	44	100	94
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	71	100	95
RASA1	Parkes Weber syndrome, 608355 Capillary malformation-arteriovenous malformation, 608354 Basal cell carcinoma, somatic, 605462	139150	70	99	96
RAX2	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381	610362	48	100	100
RB1	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200	614041	53	97	90
RB1CC1	Breast cancer, somatic, 114480	606837	55	100	95
RBBP8	Pancreatic carcinoma, somatic Seckel syndrome 2, 606744 Jawad syndrome, 251255	604124	64	100	94
RBM10	TARP syndrome, 311900	300080	42	98	88
RBM20	Cardiomyopathy, dilated, 1DD, 613172	613171	90	100	97
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	87	100	100
RBM8A	Thrombocytopenia-absent radius syndrome, 274000	605313	35	93	68
RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 Microphthalmia, isolated, with coloboma 10, 616428	180250	87	100	100
RBPJ	Adams-Oliver syndrome 3, 614814	147183	61	92	83
RD3	Leber congenital amaurosis 12, 610612	180040	100	100	100
RDH12	Leber congenital amaurosis 13, 612712	608830	80	100	100
RDH5	Fundus albipunctatus, 136880	601617	98	100	100
RDX	Deafness, autosomal recessive 24, 611022	179410	51	100	93
RECQL4	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600	603780	72	100	99
REEP1	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751	609139	73	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RELN	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436	600514	91	100	98
REN	[Hyperproreninemia] Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092	179820	80	100	100
RET	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 Renal agenesis, 191830 {Hirschsprung disease, susceptibility to, 1}, 142623	164761	96	100	99
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	62	100	98
RFTN2	No OMIM phenotype	NA	48	99	92
RFX5	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920	601863	61	100	98
RFX6	Mitchell-Riley syndrome, 615710	612659	88	100	99
RFXANK	MHC class II deficiency, complementation group B, 209920	603200	76	100	100
RFXAP	Bare lymphocyte syndrome, type II, complementation group D, 209920	601861	72	100	98
RGR	Retinitis pigmentosa 44, 613769	600342	94	100	99
RGS9	Bradyopsia, 608415	604067	67	100	99
RGS9BP	Bradyopsia, 608415	607814	58	100	100
RHAG	Anemia, hemolytic, Rh-null, regulator type, 268150 Rh-mod syndrome Overhydrated hereditary stomatocytosis, 185000	180297	102	100	95
RHBDF2	Tylosis with esophageal cancer, 148500	614404	57	100	98
RHCE	[Blood group, Rhesus], 111690 Rh-null disease, amorph type	111700	68	97	91
RHO	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880	180380	117	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	73	100	96
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075	610222	84	100	100
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650	605706	78	100	100
RIT1	Noonan syndrome 8, 615355	609591	54	100	98
RLBP1	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476 Bothnia retinal dystrophy, 607475	180090	90	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	82	100	100
RMRP	Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460 Anauxetic dysplasia, 607095	157660	No coverage data		
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	89	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	50	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	112	100	100
RNASEL	Prostate cancer 1, 601518	180435	78	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	54	100	95
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	57	100	97
RNF139	Renal cell carcinoma, 144700	603046	70	100	99
RNF145	No OMIM phenotype	NA	29	86	54
RNF168	RIDDLE syndrome, 611943	612688	99	100	100
RNF170	Ataxia, sensory, 1, autosomal dominant, 608984	614649	58	100	99
RNF212	Recombination rate QTL 1, 612042	612041	74	100	100
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	609948	54	98	90
RNF6	Esophageal carcinoma, somatic, 133239	604242	89	100	100
ROBO2	Vesicoureteral reflux 2, 610878	602431	86	99	96
ROBO3	Gaze palsy, horizontal, with progressive scoliosis, 607313	608630	71	100	99
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	63	100	98
ROM1	Retinitis pigmentosa 7, digenic, 608133	180721	65	100	100
ROR2	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310	602337	98	100	100
RP1	Retinitis pigmentosa 1, 180100	603937	61	100	96
RP1L1	Occult macular dystrophy, 613587	608581	81	100	100
RP2	Retinitis pigmentosa 2, 312600	300757	47	100	96
RPE65	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794	180069	77	100	97
RPGR	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020	312610	30	71	64
RPGRIP1	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194	605446	86	100	99
RPGRIP1L	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360	610937	77	97	95
RPIA	?Ribose 5-phosphate isomerase deficiency, 608611	180430	82	100	99
RPL11	Diamond-Blackfan anemia 7, 612562	604175	85	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RPL35A	Diamond-Blackfan anemia 5, 612528	180468	55	93	93
RPL5	Diamond-Blackfan anemia 6, 612561	603634	22	64	40
RPS10	Diamond-Blackfan anemia 9, 613308	603632	35	83	63
RPS14	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550	130620	46	90	80
RPS17	Diamond-Blackfan anemia 4, 612527	180472	14	55	27
RPS19	Diamond-Blackfan anemia 1, 105650	603474	44	91	65
RPS24	Diamond-blackfan anemia 3, 610629	602412	72	87	78
RPS26	Diamond-Blackfan anemia 10, 613309	603701	31	76	63
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844	300075	40	96	81
RPS7	Diamond-Blackfan anemia 8, 612563	603658	28	62	48
RPSA	Asplenia, isolated congenital, 271400	150370	26	98	62
RRAS2	Ovarian carcinoma	600098	28	90	53
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075	604712	87	100	100
RS1	Retinoschisis, 312700	300839	40	100	94
RSPH1	Ciliary dyskinesia, primary, 24, 615481	609314	54	100	94
RSPH4A	Ciliary dyskinesia, primary, 11, 612649	612647	78	100	100
RSPH9	Ciliary dyskinesia, primary, 12, 612650	612648	100	100	100
RSPO1	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644	609595	67	100	100
RSPO4	Anonychia congenita, 206800	610573	79	100	98
RTEL1	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	58	97	87
RTN2	Spastic paraplegia 12, autosomal dominant, 604805	603183	77	100	100
RTTN	Polymicrogyria with seizures, 614833	610436	71	99	96
RUNX1	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399	151385	50	100	96
RUNX2	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510	600211	99	100	100
RXFP2	No OMIM phenotype	606655	79	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RYR1	{Malignant hyperthermia susceptibility 1}, 145600 Central core disease, 117000 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 King-Denborough syndrome, 145600	180901	79	98	96
RYR2	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996	180902	83	100	99
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550	604490	68	100	99
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	79	100	100
SALL1	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480	602218	99	100	98
SALL4	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750	607343	101	100	98
SAMD9	Tumoral calcinosis, familial, normophosphatemic, 610455	610456	76	100	98
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	76	100	94
SAR1B	Chylomicron retention disease, 246700	607690	84	100	89
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845	612804	65	100	99
SART3	No OMIM phenotype	611684	82	100	100
SAT1	No OMIM phenotype	313020	53	100	98
SATB2	Glass syndrome, 612313	608148	67	100	99
SBDS	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135	607444	61	100	96
SBF2	Charcot-Marie-Tooth disease, type 4B2, 604563	607697	71	100	97
SC5D	Lathosterolosis, 607330	602286	86	100	100
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	602257	88	100	100
SCARF2	Van den Ende-Gupta syndrome, 600920	613619	59	98	87
SCN10A	Episodic pain syndrome, familial, 2, 615551	604427	107	100	100
SCN11A	Neuropathy, hereditary sensory and autonomic, type VII, 615548 Episodic pain syndrome, familial, 3, 615552	604385	81	100	100
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403	182389	100	100	99
SCN1B	Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377	600235	102	97	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SCN2A	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721	182390	100	100	99
SCN2B	Atrial fibrillation, familial, 14, 615378	601327	121	100	95
SCN3B	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120	608214	70	100	100
SCN4A	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345	603967	120	100	100
SCN4B	Long QT syndrome-10, 611819 Atrial fibrillation, familial, 17, 611819	608256	63	100	100
SCN5A	Long QT syndrome-3, 603830 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900 Heart block, nonprogressive, 113900 Ventricular fibrillation, familial, 1, 603829 Sick sinus syndrome 1, 608567 Cardiomyopathy, dilated, 1E, 601154 {Sudden infant death syndrome, susceptibility to}, 272120 Atrial fibrillation, familial, 10, 614022	600163	102	100	100
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558	600702	97	100	99
SCN9A	Erythermalgia, primary, 133020 Paroxysmal extreme pain disorder, 167400, Insensitivity to pain, congenital, 243000 Febrile seizures, familial, 3B, 613863 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208 HSAN2D, autosomal recessive, 243000	603415	98	100	98
SCNN1A	Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 2, 613021	600228	92	100	100
SCNN1B	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 1, 211400	600760	97	100	100
SCNN1G	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071	600761	103	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SCO1	Mitochondrial complex IV deficiency, 220110	603644	69	100	98
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	94	100	100
SCP2	Leukoencephalopathy with dystonia and motor neuropathy, 613724	184755	69	100	93
SDCCAG8	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993	613524	70	100	99
SDHA	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165	600857	43	77	64
SDHAF1	Mitochondrial complex II deficiency, 252011	612848	45	100	100
SDHAF2	Paragangliomas 2, 601650	613019	91	100	100
SDHB	Paragangliomas 4, 115310 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764	185470	75	100	99
SDHC	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764	602413	73	100	100
SDHD	Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300 Carcinoid tumors, intestinal, 114900 Merkel cell carcinoma, somatic Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 3, 615106 Mitochondrial complex II deficiency, 252011	602690	71	100	100
SEC23A	Craniolenticulosutural dysplasia, 607812	610511	69	100	94
SEC23B	Dyserythropoietic anemia, congenital, type II, 224100	610512	79	100	97
SEC63	Polycystic liver disease, 174050	608648	55	100	90
SECISBP2	Thyroid hormone metabolism, abnormal, 609698	607693	79	100	97
SEMA3E	?CHARGE syndrome, 214800	608166	72	100	100
SEMA4A	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283	607292	91	100	100
SEPN1	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310	606210	78	88	84
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	92	100	100
SEPT12	Spermatogenic failure 10, 614822	611562	80	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SEPT9	Leukemia, acute myeloid, therapy-related Ovarian carcinoma Amyotrophy, hereditary neuralgic, 162100	604061	71	100	99
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	69	100	99
SERPINA1	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	107400	100	100	100
SERPINA3	Alpha-1-antichymotrypsin deficiency Cerebrovascular disease, occlusive	107280	123	100	100
SERPINA6	Corticosteroid-binding globulin deficiency, 611489	122500	96	100	100
SERPINA7	Thyroxine-binding globulin deficiency	314200	52	100	100
SERPINB6	?Deafness, autosomal recessive 91, 613453	173321	99	100	100
SERPINB7	Palmoplantar keratoderma, Nagashima type, 615598	603357	40	98	80
SERPINC1	Thrombophilia due to antithrombin III deficiency, 613118	107300	87	100	100
SERPIND1	Thrombophilia due to heparin cofactor II deficiency, 612356	142360	67	100	100
SERPINE1	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}	173360	113	100	100
SERPINF1	Osteogenesis imperfecta, type VI, 613982	172860	94	100	100
SERPINF2	Alpha-2-plasmin inhibitor deficiency, 262850	613168	100	100	100
SERPING1	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790	606860	88	100	100
SERPINH1	{Preterm premature rupture of the membranes, susceptibility to}, 610504 ?Osteogenesis imperfecta, type X, 613848	600943	91	100	100
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218	602445	71	100	100
SETBP1	Schinzel-Giedion midface retraction syndrome, 269150 Mental retardation, autosomal dominant 29, 616078	611060	74	98	97
SETD1A	No OMIM phenotype	611052	57	97	91
SETD5	Mental retardation, autosomal dominant 23, 615761	615743	57	99	96
SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002	608465	76	100	99
SF3B1	Myelodysplastic syndrome, somatic, 614286	605590	76	100	100
SF3B4	Acrofacial dysostosis 1, Nager type, 154400	605593	34	96	73
SFTPA2	Pulmonary fibrosis, idiopathic, 178500	178642	68	100	100
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1, 265120	178640	66	100	100
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2, 610913	178620	84	100	100
SFXN4	Combined oxidative phosphorylation deficiency 18, 615578	615564	46	97	86
SGCA	Muscular dystrophy, limb-girdle, type 2D, 608099	600119	89	100	100
SGCB	Muscular dystrophy, limb-girdle, type 2E, 604286	600900	107	96	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SGCD	Muscular dystrophy, limb-girdle, type 2F, 601287 Cardiomyopathy, dilated, 1L, 606685	601411	78	100	100
SGCE	Dystonia-11, myoclonic, 159900	604149	64	94	93
SGCG	Muscular dystrophy, limb-girdle, type 2C, 253700	608896	67	100	100
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900	605270	86	94	94
SH2B3	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100	605093	62	96	84
SH2D1A	Lymphoproliferative syndrome, X-linked, 1, 308240	300490	45	100	97
SH3BP2	Cherubism, 118400	602104	71	91	91
SH3PXD2B	Frank-ter Haar syndrome, 249420	613293	87	100	100
SH3TC2	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353	608206	73	100	100
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	56	93	82
SHH	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160	600725	80	100	97
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	78	100	100
SHOX	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700	312865	58	96	87
SHROOM4	?Stocco dos Santos X-linked mental retardation syndrome, 300434	300579	44	100	93
SI	Sucrase-isomaltase deficiency, congenital, 222900	609845	63	98	94
SIGMAR1	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726	601978	56	100	100
SIL1	Marinesco-Sjogren syndrome, 248800	608005	85	100	100
SIM1	Obesity, severe, 601665	603128	85	100	99
SIX1	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192	601205	90	100	100
SIX3	Holoprosencephaly-2, 157170 Schizencephaly, 269160	603714	103	100	100
SIX5	Branchiootorenal syndrome 2, 610896	600963	45	94	82
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, 212550	606326	108	100	100
SKI	Shprintzen-Goldberg syndrome, 182212	164780	55	99	97
SKIV2L	Trichohepatoenteric syndrome 2, 614602	600478	60	99	91
SLC10A2	Bile acid malabsorption, primary, 613291	601295	94	100	100
SLC11A2	Anemia, hypochromic microcytic, 206100	600523	71	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC12A1	Bartter syndrome, type 1, 601678	600839	83	100	99
SLC12A3	Gitelman syndrome, 263800	600968	84	100	100
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	71	100	99
SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095	600682	76	100	100
SLC16A12	Cataract, juvenile, with microcornea and glucosuria, 612018	611910	72	100	99
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	42	99	89
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	75	100	99
SLC17A8	Deafness, autosomal dominant 25, 605583	607557	81	100	99
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270	603941	90	100	100
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	118	100	100
SLC1A3	Episodic ataxia, type 6, 612656	600111	92	100	100
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600	158378	83	100	97
SLC22A12	Hypouricemia, renal, 220150	607096	72	100	100
SLC22A18	Breast cancer, somatic, 114480 Rhabdomyosarcoma, somatic, 268210 Lung cancer, somatic, 211980	602631	65	96	88
SLC22A5	Carnitine deficiency, systemic primary, 212140	603377	97	100	100
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830	603617	85	100	100
SLC24A5	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750	609802	56	100	100
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182	190315	40	99	90
SLC25A12	Hypomyelination, global cerebral, 612949	603667	87	100	100
SLC25A13	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814	603859	70	100	96
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	106	90	86
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	85	100	100
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138	613698	101	100	100
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	63	100	100
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773	600370	70	97	93
SLC25A38	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950	610819	40	92	75
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418	103220	91	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC26A2	Diastrophic dysplasia, 222600 Atelosteogenesis II, 256050 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 De la Chapelle dysplasia, 256050	606718	102	100	100
SLC26A3	Diarrhea 1, secretory chloride, congenital, 214700	126650	78	100	100
SLC26A4	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791	605646	82	100	98
SLC26A5	?Deafness, autosomal recessive 61, 613865	604943	90	100	100
SLC26A8	Spermatogenic failure 3, 606766	608480	44	99	88
SLC27A4	Ichthyosis prematurity syndrome, 608649	604194	93	100	100
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, 602782	612373	98	100	100
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 Dystonia 9, 601042 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	104	100	100
SLC2A10	Arterial tortuosity syndrome, 208050	606145	95	100	100
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi-Bickel syndrome, 227810	138160	90	100	100
SLC2A9	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076	606142	73	100	94
SLC30A10	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280	611146	103	100	100
SLC30A2	Zinc deficiency, transient neonatal, 608118	609617	76	100	100
SLC33A1	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482	603690	62	100	96
SLC34A1	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Fanconi renotubular syndrome 2, 613388	182309	79	100	100
SLC34A2	Pulmonary alveolar microlithiasis, 265100 ?Testicular microlithiasis, 610441	604217	81	100	98
SLC34A3	Hypophosphatemic rickets with hypercalciuria, 241530	609826	82	100	99
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	76	100	100
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	29	98	80
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	103	100	100
SLC35D1	Schneckenbecken dysplasia, 269250	610804	72	100	91
SLC36A2	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500	608331	73	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	80	100	96
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	615585	29	94	75
SLC39A13	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350	608735	78	100	100
SLC39A4	Acrodermatitis enteropathica, 201100	607059	62	100	98
SLC3A1	Cystinuria, 220100	104614	99	100	100
SLC40A1	Hemochromatosis, type 4, 606069	604653	80	100	100
SLC45A2	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240	606202	78	100	99
SLC46A1	Folate malabsorption, hereditary, 229050	611672	71	100	100
SLC4A1	Ovalocytosis, SA type, 166900 Spherocytosis, type 4, 612653 [Malaria, resistance to], 611162 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 [Blood group, Diego], 110500 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Blood group, Froese], 601551 [Blood group, Swann], 601550 Cryohydrocytosis, 185020	109270	78	100	100
SLC4A11	Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal dystrophy, Fuchs endothelial, 4, 613268	610206	79	100	97
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	71	100	99
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500	613350	60	100	100
SLC5A1	Glucose/galactose malabsorption, 606824	182380	94	100	100
SLC5A2	Renal glucosuria, 233100	182381	72	100	100
SLC5A5	Thyroid dyshormonogenesis 1, 274400	601843	62	100	97
SLC5A7	Neuronopathy, distal hereditary motor, type VIIA, 158580	608761	75	100	100
SLC6A19	Hartnup disorder, 234500 Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500	608893	92	100	100
SLC6A2	Orthostatic intolerance, 604715	163970	97	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC6A20	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	605616	87	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	87	100	100
SLC6A5	Hyperekplexia 3, 614618	604159	87	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	19	81	42
SLC7A14	Retinitis pigmentosa 68, 615725	615720	55	100	96
SLC7A7	Lysinuric protein intolerance, 222700	603593	79	100	98
SLC7A9	Cystinuria, 220100	604144	98	100	100
SLC9A3R1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287	604990	86	100	98
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243	300231	50	100	96
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic, 237450	604843	52	100	95
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic, 237450	605495	45	99	94
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441	601460	78	100	100
SLITRK1	Tourette syndrome, 137580 ?Trichotillomania, 613229	609678	62	100	100
SLITRK6	Deafness and myopia, 221200	609681	93	100	100
SLURP1	Meleda disease, 248300	606119	77	100	100
SLX4	Fanconi anemia, complementation group P, 613951	613278	82	100	100
SMAD3	Loeys-Dietz syndrome 3, 613795	603109	93	100	100
SMAD4	Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900	600993	76	100	98
SMAD6	Aortic valve disease 2, 614823	602931	71	94	84
SMAD9	Pulmonary hypertension, primary, 2, 615342	603295	78	100	100
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	78	98	96
SMARCA4	{Rhabdoid tumor predisposition syndrome 2}, 613325 Mental retardation, autosomal dominant 16, 614609	603254	91	100	100
SMARCAD1	Adermatoglyphia, 136000	612761	60	100	95
SMARCAL1	Schimke immunoosseous dysplasia, 242900	606622	72	100	100
SMARCB1	Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 Mental retardation, autosomal dominant 15, 614608 {Schwannomatosis-1, susceptibility to}, 162091	601607	123	100	100
SMC1A	Cornelia de Lange syndrome 2, 300590	300040	48	100	91
SMC3	Cornelia de Lange syndrome 3, 610759	606062	50	93	84
SMCHD1	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901	614982	62	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SMN1	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150	600354	34	97	78
SMO	Basal cell carcinoma, somatic	601500	86	100	94
SMOC1	Microphthalmia with limb anomalies, 206920	608488	77	100	100
SMOC2	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400	607223	71	100	94
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	110	100	100
SMPX	Deafness, X-linked 4, 300066	300226	50	100	94
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583	300105	39	95	94
SNAI2	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800	602150	71	100	92
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	99	100	100
SNCA	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601	163890	82	100	100
SNCB	Dementia, Lewy body, 127750	602569	70	100	100
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	88	100	100
SNRNP200	Retinitis pigmentosa 33, 610359	601664	105	100	100
SNRPE	Hypotrichosis 11, 615059	128260	27	89	59
SNRPN	Prader-Willi syndrome, 176270	182279	90	100	98
SNTA1	Long QT syndrome 12, 612955	601017	60	89	77
SNX10	Osteopetrosis, autosomal recessive 8, 615085	614780	72	100	100
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	79	98	94
SOD1	Amyotrophic lateral sclerosis 1, 105400	147450	51	100	100
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	65	100	95
SOST	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860	605740	79	100	100
SOX10	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136	602229	45	99	87
SOX17	Vesicoureteral reflux 3, 613674	610928	58	100	91
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940	601618	31	83	55
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	113	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000	313430	34	99	76
SOX9	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290	608160	73	98	91
SP110	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948	604457	89	100	99
SP7	?Osteogenesis imperfecta, type XII, 613849	606633	89	100	98
SPAG1	Ciliary dyskinesia, primary, 28, 615505	603395	36	89	69
SPAST	Spastic paraplegia 4, autosomal dominant, 182601	604277	49	100	91
SPATA16	?Spermatogenic failure 6, 102530	609856	76	100	100
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232	609868	71	100	96
SPECC1L	?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410	614140	65	100	97
SPG11	Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668	610844	73	100	99
SPG20	Troyer syndrome, 275900	607111	71	100	99
SPG21	Mast syndrome, 248900	608181	85	100	99
SPG7	Spastic paraplegia 7, autosomal recessive, 607259	602783	93	98	92
SPINK1	Pancreatitis, hereditary, 167800 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Tropical calcific pancreatitis, 608189	167790	47	100	100
SPINK5	Netherton syndrome, 256500 Atopy, 147050	605010	81	100	96
SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420	605124	61	100	90
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	98	100	94
SPRED1	Legius syndrome, 611431	609291	67	100	95
SPRY4	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266	607984	80	100	100
SPTA1	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970	182860	89	100	99
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	87	100	100
SPTB	Elliptocytosis-3 Spherocytosis, type 2, 616649 Anemia, neonatal hemolytic, fatal and near-fatal	182870	90	100	100
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386	604985	82	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA, 162400	605712	70	100	97
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640	605713	93	100	100
SQSTM1	Paget disease of bone 3, 167250 Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437	601530	96	100	100
SRC	Colon cancer, advanced, somatic	190090	78	100	99
SRCAP	Floating-Harbor syndrome, 136140	611421	101	100	99
SRD5A2	Pseudovaginal perineoscrotal hypospadias, 264600	607306	67	100	100
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	105	100	100
SRP72	Bone marrow failure syndrome 1, 614675	602122	48	99	89
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	50	99	91
SRY	46XY sex reversal 1, 400044 46XX sex reversal 1, 400045	480000	92	100	100
SSTR5	Somatostatin analog, resistance to	182455	111	100	100
ST14	Ichthyosis, congenital, autosomal recessive 11, 602400	606797	81	100	97
ST3GAL3	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006	606494	111	100	100
ST3GAL5	Amish infantile epilepsy syndrome, 609056	604402	85	93	93
STAC3	Native American myopathy, 255995	615521	44	100	93
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	37	96	79
STAR	Lipoid adrenal hyperplasia, 201710	600617	88	100	100
STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162	600555	76	100	96
STAT3	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 615952	102582	81	100	98
STAT5B	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590	604260	69	99	90
STIL	Microcephaly 7, primary, autosomal recessive, 612703	181590	81	100	97
STIM1	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070	605921	82	100	99
STK11	Peutz-Jeghers syndrome, 175200 Melanoma, malignant, somatic Pancreatic cancer, 260350 Testicular tumor, somatic, 273300	602216	64	100	98
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868	604965	77	100	98
STOX1	Preeclampsia/eclampsia 4, 609404	609397	74	89	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
STRA6	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186	610745	58	100	100
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	83	100	100
STRC	Deafness, autosomal recessive 16, 603720	606440	73	100	97
STS	Ichthyosis, X-linked, 308100	300747	67	100	99
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	92	100	100
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, 613101	601717	85	100	99
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	38	86	75
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	74	100	98
SUFU	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174 Basal cell nevus syndrome, 109400	607035	84	100	100
SUMF1	Multiple sulfatase deficiency, 272200	607939	87	100	100
SUMO1	Orofacial cleft 10, 613705	601912	11	59	12
SUOX	Sulfite oxidase deficiency, 272300	606887	104	100	100
SURF1	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684	185620	71	88	88
SYCP3	Spermatogenic failure 4, 270960 Pregnancy loss, recurrent, 4, 270960	604759	64	100	85
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491	313440	37	86	67
SYNE1	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998	608441	87	100	100
SYNE2	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999	608442	73	100	96
SYNE4	Deafness, autosomal recessive 76, 615540	615535	36	99	85
SYNGAP1	Mental retardation, autosomal dominant 5, 612621	603384	55	95	82
SYNJ1	Parkinson disease 20, early-onset, 615530	604297	42	96	79
SYP	Mental retardation, X-linked 96, 300802	313475	48	100	100
SYT14	Spinocerebellar ataxia, autosomal recessive 11, 614229	610949	92	93	89
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	57	99	93
T	{Neural tube defects, susceptibility to}, 182940 Sacral agenesis with vertebral anomalies, 615709	601397	97	100	99
TAB2	Congenital heart defects, nonsyndromic, 2, 614980	605101	91	100	100
TAC3	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839	162330	69	100	91
TACR3	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840	162332	75	100	100
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870	137290	107	100	100
TAF1	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966	313650	78	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TAF2	Mental retardation, autosomal recessive 40, 615599	604912	63	100	99
TAL1	Leukemia, T-cell acute lymphocytic, somatic, 613065	187040	40	85	62
TAL2	Leukemia, T-cell acute lymphocytic, somatic, 613065	186855	55	100	100
TALDO1	Transaldolase deficiency, 606003	602063	96	100	98
TAP1	Bare lymphocyte syndrome, type I, 604571	170260	46	95	77
TAP2	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis	170261	43	99	87
TAPBP	Bare lymphocyte syndrome, type I, 604571	601962	53	100	96
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069	605078	75	100	100
TAT	Tyrosinemia, type II, 276600	613018	82	100	100
TAZ	Barth syndrome, 302060	300394	64	100	98
TBC1D20	Warburg micro syndrome 4, 615663	611663	51	93	85
TBC1D24	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338 DOOR syndrome, 220500 Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044	613577	92	100	100
TBCE	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410	604934	80	99	91
TBP	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600	600075	96	100	100
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Velocardiofacial syndrome, 192430 Tetralogy of Fallot, 187500	602054	55	79	66
TBX15	Cousin syndrome, 260660	604127	69	100	97
TBX19	Adrenocorticotropic hormone deficiency, 201400	604614	116	100	100
TBX20	Atrial septal defect 4, 611363	606061	70	100	94
TBX21	{Asthma, aspirin-induced, susceptibility to}, 208550 Asthma and nasal polyps, 208550	604895	82	97	88
TBX22	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905	300307	50	100	96
TBX3	Ulnar-mammary syndrome, 181450	601621	59	100	97
TBX4	Ischiocoxopodopatellar syndrome, 147891	601719	100	100	100
TBX5	Holt-Oram syndrome, 142900	601620	72	100	100
TBXAS1	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158	274180	96	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TCAP	Muscular dystrophy, limb-girdle, type 2G, 601954 Cardiomyopathy, hypertrophic, 25, 607487	604488	55	100	100
TCF12	Craniosynostosis 3, 615314	600480	51	100	94
TCF4	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267	602272	73	100	98
TCIRG1	Osteopetrosis, autosomal recessive 1, 259700	604592	77	96	92
TCN2	Transcobalamin II deficiency, 275350	613441	110	100	100
TCOF1	Treacher Collins syndrome 1, 154500	606847	78	100	98
TCTN1	Joubert syndrome 13, 614173	609863	70	100	97
TCTN2	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654	613846	93	100	100
TCTN3	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815	613847	60	100	96
TDGF1	Forebrain defects	187395	53	92	76
TDP1	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250	607198	77	100	96
TDRD7	Cataract 36, 613887	611258	91	100	99
TEAD1	Sveinsson choreoretinal atrophy, 108985	189967	74	100	100
TECPR2	Spastic paraplegia 49, autosomal recessive, 615031	615000	92	100	100
TECR	Mental retardation, autosomal recessive 14, 614020	610057	68	99	94
TECTA	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629	602574	101	100	100
TEK	Venous malformations, multiple cutaneous and mucosal, 600195	600221	92	100	100
TENM3	?Microphthalmia, isolated, with coloboma 9, 615145	610083	62	98	94
TERC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743	602322	No coverage data		
TET2	Myelodysplastic syndrome, somatic, 614286	612839	82	100	100
TEX28	No OMIM phenotype	300092	0	0	0
TF	Atransferrinemia, 209300	190000	93	100	100
TFAP2A	Branchiooculofacial syndrome, 113620	107580	66	100	96
TFAP2B	Char syndrome, 169100	601601	79	100	100
TFE3	Renal cell carcinoma, papillary, 1, 300854	314310	37	100	89
TFG	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, autosomal recessive, 615658	602498	49	93	72
TFR2	Hemochromatosis, type 3, 604250	604720	68	100	100
TG	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175	188450	88	100	98
TGFB1	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700	190180	67	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TGFB2	Loeys-Dietz syndrome 4, 614816	190220	107	100	100
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582	190230	96	100	100
TGFBI	Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820	601692	83	99	93
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	111	94	94
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	100	100	100
TGIF1	Holoprosencephaly-4, 142946	602630	97	100	100
TGM1	Ichthyosis, congenital, autosomal recessive 1, 242300	190195	98	100	100
TGM5	Peeling skin syndrome 2, 609796	603805	112	100	100
TGM6	Spinocerebellar ataxia 35, 613908	613900	82	100	100
TH	Segawa syndrome, recessive, 605407	191290	55	100	95
THAP1	Dystonia 6, torsion, 602629	609520	84	100	96
THBD	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926	188040	89	100	100
THOC6	Beaulieu-Boycott-Innes syndrome, 613680	615403	101	100	100
THPO	Thrombocythemia 1, 187950	600044	48	81	79
THRA	Hypothyroidism, congenital, nongoitrous, 6, 614450	190120	100	100	100
THRB	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650	190160	89	100	100
TIA1	Welander distal myopathy, 604454	603518	44	85	69
TIMM8A	Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150	300356	42	100	100
TIMP3	Sorsby fundus dystrophy, 136900	188826	88	100	100
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130	604319	92	100	100
TJP2	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878	607709	83	100	99
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560	188250	72	100	94
TLL1	Atrial septal defect 6, 613087	606742	77	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TLR4	Endotoxin hyporesponsiveness {Macular degeneration, age-related, 10}, 611488 {Colorectal cancer, susceptibility to}, 114500	603030	71	100	100
TMC1	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705	606706	68	100	100
TMC6	Epidermodysplasia verruciformis, 226400	605828	61	100	99
TMC8	Epidermodysplasia verruciformis, 226400	605829	80	100	95
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614123	60	100	98
TMEM126A	Optic atrophy 7, 612989	612988	70	100	96
TMEM138	Joubert syndrome 16, 614465	614459	67	100	100
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	76	100	100
TMEM181	No OMIM phenotype	613209	41	100	90
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	87	100	100
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	56	98	93
TMEM237	Joubert syndrome 14, 614424	614423	80	100	99
TMEM38B	Osteogenesis imperfecta, type XIV, 615066	611236	35	100	85
TMEM67	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Nephronophthisis 11, 613550	609884	51	99	90
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	75	100	99
TMIE	Deafness, autosomal recessive 6, 600971	607237	68	99	88
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency, 300872	300777	42	98	87
TMPRSS15	Enterokinase deficiency, 226200	606635	72	99	97
TMPRSS3	Deafness, autosomal recessive 8/10, 601072	605511	94	100	100
TMPRSS6	Iron-refractory iron deficiency anemia, 206200	609862	69	100	99
TNC	Deafness, autosomal dominant 56, 615629	187380	64	99	92
TNFRSF10B	Squamous cell carcinoma, head and neck, 275355	603612	102	100	100
TNFRSF11A	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301	603499	66	96	96
TNFRSF11B	Paget disease of bone 5, juvenile-onset, 239000	602643	131	100	100
TNFRSF13B	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500	604907	77	100	100
TNFRSF13C	Immunodeficiency, common variable, 4, 613494	606269	30	87	62

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TNFRSF1A	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810	191190	68	100	100
TNFSF11	Osteopetrosis, autosomal recessive 2, 259710	602642	65	100	98
TNNC1	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243	191040	134	100	100
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B, 601680	191043	63	100	100
TNNI3	Cardiomyopathy, hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 1, 115210 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286	191044	81	100	96
TNNT1	Nemaline myopathy 5, Amish type, 605355	191041	71	100	96
TNNT2	Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494	191045	75	100	100
TNNT3	Arthyrgryposis, distal, type 2B, 601680	600692	71	100	97
TNXB	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963	600985	96	100	98
TOP1	DNA topoisomerase I, camptothecin-resistant	126420	71	99	94
TOP2A	DNA topoisomerase II, resistance to inhibition of, by amsacrine	126430	69	100	97
TOPORS	Retinitis pigmentosa 31, 609923	609507	76	100	100
TP53	Colorectal cancer, 114500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, 114550 Osteosarcoma, 259500 Choroid plexus papilloma, 260500 Nasopharyngeal carcinoma, 607107 Pancreatic cancer, 260350 Adrenal cortical carcinoma, 202300 Breast cancer, 114480 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800	191170	70	100	100
TP63	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 ADULT syndrome, 103285 Limb-mammary syndrome, 603543 Rapp-Hodgkin syndrome, 129400 Orofacial cleft 8, 129400	603273	109	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	41	92	81
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458	606370	63	100	100
TPM1	Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878 Left ventricular noncompaction 9, 611878	191010	80	100	100
TPM2	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285	190990	79	98	91
TPM3	Nemaline myopathy 1, autosomal dominant or recessive, 609284 CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310	191030	83	100	99
TPMT	6-mercaptopurine sensitivity, 610460	187680	34	90	76
TPO	Thyroid dyshormonogenesis 2A, 274500	606765	84	99	92
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	79	100	98
TPRN	Deafness, autosomal recessive 79, 613307	613354	49	80	71
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	44	96	82
TRAPPC2	Spondyloepiphyseal dysplasia tarda, 313400	300202	31	67	44
TRAPPC9	Mental retardation, autosomal recessive 13, 613192	611966	85	100	99
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441	603283	52	95	82
TREM2	Nasu-Hakola disease, 221770	605086	79	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700	606609	129	100	100
TRHR	Thyrotropin-releasing hormone resistance, generalized	188545	85	100	100
TRIM24	No OMIM phenotype	603406	78	99	94
TRIM32	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988	602290	77	100	100
TRIM33	No OMIM phenotype	605769	74	97	91
TRIM37	Mulibrey nanism, 253250	605073	74	100	97
TRIOBP	Deafness, autosomal recessive 28, 609823	609761	120	100	99
TRIP11	Achondrogenesis, type IA, 200600	604505	57	97	91
TRMU	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070	610230	76	100	100
TRPA1	Episodic pain syndrome, familial, 615040	604775	53	96	85
TRPC6	Glomerulosclerosis, focal segmental, 2, 603965	603652	75	94	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216	603576	87	100	100
TRPM4	Progressive familial heart block, type IB, 604559	606936	77	100	98
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	81	100	99
TRPS1	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351	604386	70	100	100
TRPV3	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400	607066	92	100	96
TRPV4	Brachyolmia type 3, 113500 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Metatropic dysplasia, 156530 Hereditary motor and sensory neuropathy, type IIc, 606071 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level QTL 1], 613508 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Digital arthropathy-brachydactyly, familial, 606835	605427	92	100	100
TSC1	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341	605284	94	100	99
TSC2	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690	191092	94	100	100
TSEN2	Pontocerebellar hypoplasia type 2B, 612389	608753	84	100	98
TSEN34	Pontocerebellar hypoplasia type 2C, 612390	608754	56	90	83
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	96	96
TSFM	Combined oxidative phosphorylation deficiency 3, 610505	604723	59	100	98
TSG101	Breast cancer, somatic, 114480	601387	54	100	96
TSHB	Hypothryoidism, congenital, nongoitrous 4, 275100	188540	120	100	100
TSHR	Hypothyroidism, congenital, nongoitrous, 1 275200 Thyroid adenoma, hyperfunctioning, somatic Hyperthyroidism, nonautoimmune, 609152 Thyroid carcinoma with thyrotoxicosis Hyperthyroidism, familial gestational, 603373	603372	93	100	98
TSHZ1	Aural atresia, congenital, 607842	614427	90	99	99
TSPAN12	Exudative vitreoretinopathy 5, 613310	613138	85	100	100
TSPAN7	Mental retardation, X-linked 58, 300210	300096	40	100	85
TSPEAR	Deafness, autosomal recessive 98, 614861	612920	97	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome, 608800	604714	99	100	100
TTBK2	Spinocerebellar ataxia 11, 604432	611695	70	100	99
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	57	90	78
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	612014	70	100	97
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	77	100	98
TTC7A	Gastrointestinal defects and immunodeficiency syndrome, 243150	609332	54	99	90
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	64	100	90
TTI2	Mental retardation, autosomal recessive 39, 615541	614426	79	100	100
TTN	Cardiomyopathy, familial hypertrophic, 9, 613765 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Myopathy, early-onset, with fatal cardiomyopathy, 611705	188840	90	100	99
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	70	96	89
TTR	Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430	176300	87	100	100
TUBA1A	Lissencephaly 3, 611603	602529	34	93	78
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180	605742	116	100	99
TUBB1	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112	612901	104	100	100
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	51	100	89
TUBB2B	Polymicrogyria, symmetric or asymmetric, 610031	612850	65	100	94
TUBB3	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039	602661	75	100	100
TUBB4A	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	60	97	93
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	65	100	99
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270	610053	94	100	100
TUFM	Combined oxidative phosphorylation deficiency 4, 610678	602389	76	100	97
TULP1	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843	602280	65	100	97
TUSC3	Mental retardation, autosomal recessive 7, 611093	601385	74	100	100
TWIST1	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750	601622	79	100	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TWIST2	Focal facial dermal dysplasia 3, Setleis type, 227260 Barber-Say syndrome, 209885 Ablepharon-macrostomia syndrome, 200110	607556	103	100	100
TYK2	Immunodeficiency 35, 611521	176941	73	100	100
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	131222	54	99	84
TYR	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800	606933	71	100	100
TYROBP	Nasu-Hakola disease, 221770	604142	44	100	92
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	107	100	100
UBA1	Spinal muscular atrophy, X-linked 2, infantile, 301830	314370	63	100	98
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860	312180	33	100	96
UBE3A	Angelman syndrome, 105830	601623	61	100	99
UBE3B	Kaufman oculocerebrofacial syndrome, 244450	608047	49	99	90
UBIAD1	Corneal dystrophy, Schnyder type, 121800	611632	83	100	100
UBQLN2	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857	300264	47	100	100
UBR1	Johanson-Blizzard syndrome, 243800	605981	69	100	95
UGT1A1	Crigler-Najjar syndrome, type I, 218800 [Gilbert syndrome], 143500 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816	191740	100	100	100
UMOD	Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860 Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886	191845	82	100	99
UMPS	Orotic aciduria, 258900	613891	105	100	100
UNG	Immunodeficiency with hyper IgM, type 5, 608106	191525	79	100	100
UPB1	Beta-ureidopropionase deficiency, 613161	606673	108	100	100
UPF3B	Mental retardation, X-linked, syndromic 14, 300676	300298	32	99	83
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158	191330	81	100	89
UQCRC2	Mitochondrial complex III deficiency, nuclear type 5, 615160	191329	42	98	81
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	167	100	100
UROC1	?Urocanase deficiency, 276880	613012	82	100	100
UROD	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100	613521	85	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
UROS	Porphyria, congenital erythropoietic, 263700	606938	60	100	100
USB1	Poikiloderma with neutropenia, 604173	613276	85	100	100
USH1C	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092	605242	70	100	91
USH1G	Usher syndrome, type 1G, 606943	607696	86	100	99
USH2A	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809	608400	90	100	100
USP9Y	Spermatogenic failure, Y-linked, 2, 415000	400005	42	98	86
UTP4	Cirrhosis, North American Indian childhood type, 604901	607456	No coverage data		
UVSSA	UV-sensitive syndrome 3, 614640	614632	81	100	99
VANGL1	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940	610132	89	100	100
VANGL2	Neural tube defects, 182940	600533	83	100	99
VAPB	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980	605704	85	100	89
VAX1	?Microphthalmia, syndromic 11, 614402	604294	54	90	84
VCAN	Wagner syndrome 1, 143200	118661	78	100	100
VCL	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255	193065	87	100	100
VCP	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687	601023	84	100	99
VDR	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710	601769	90	100	100
VHL	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400	608537	75	98	87
VIM	?Cataract 30, pulverulent, 116300	193060	96	100	100
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404	613401	69	100	94
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700	608547	107	100	100
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	105	100	99
VPS13A	Choreoacanthocytosis, 200150	605978	57	99	94
VPS13B	Cohen syndrome, 216550	607817	83	100	99
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085	608552	83	100	99
VPS35	{Parkinson disease 17}, 614203	601501	52	89	79
VPS37A	Spastic paraplegia 53, autosomal recessive, 614898	609927	46	76	55

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
VPS45	Neutropenia, severe congenital, 5, autosomal recessive, 615285	610035	48	97	87
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	78	100	96
VSX1	Keratoconus 1, 148300 Corneal dystrophy, posterior polymorphous, 1, 122000 Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195	605020	62	89	78
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	74	100	100
VWF	von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 1, 193400 von Willibrand disease, type 3, 277480	613160	65	99	93
WAS	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900	300392	35	94	73
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	67	100	93
WDR11	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858	606417	75	98	95
WDR19	Nephronophthisis 13, 614377 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Senior-Loken syndrome 8, 616307	608151	73	100	98
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	613363	48	100	93
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613602	82	100	95
WDR36	Glaucoma 1, open angle, G, 609887	609669	72	100	96
WDR45	Neurodegeneration with brain iron accululation 5, 300894	300526	32	89	78
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	615462	41	94	81
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317	613583	90	100	100
WDR72	Amelogenesis imperfecta, type IIA3, 613211	613214	77	100	99
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185	614218	90	100	100
WFS1	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 ?Cataract 41, 116400	606201	102	100	100
WHRN	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383	607928	No coverage data		
WHSC1L1	Leukemia, acute myeloid, 601626	607083	39	98	86
WIPF1	?Wiskott-Aldrich syndrome 2, 614493	602357	55	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230	603400	85	100	100
WNK1	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300	605232	83	100	98
WNK4	Pseudohypoaldosteronism, type IIB, 614491	601844	89	100	100
WNT1	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221	164820	98	100	96
WNT10A	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400	606268	90	100	99
WNT10B	Split-hand/foot malformation 6, 225300	601906	91	100	100
WNT3	?Tetra-amelia syndrome, 273395	165330	86	100	95
WNT4	SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330	603490	125	96	92
WNT5A	Robinow syndrome, autosomal dominant 1, 180700	164975	117	100	99
WNT7A	Ulna and fibula, absence of, with severe limb deficiency, 276820 Fuhrmann syndrome, 228930	601570	103	100	100
WRAP53	Dyskeratosis congenita, autosomal recessive 3, 613988	612661	97	100	99
WRN	Werner syndrome, 277700	604611	71	100	92
WT1	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240	607102	61	99	90
WWOX	Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211	605131	72	100	100
XDH	Xanthinuria, type I, 278300	607633	80	100	99
XIAP	Lymphoproliferative syndrome, X-linked, 2, 300635	300079	36	99	87
XK	McLeod syndrome with or without chronic granulomatous disease, 300842	314850	54	100	98
XPA	Xeroderma pigmentosum, group A, 278700	611153	47	98	86
XPC	Xeroderma pigmentosum, group C, 278720	613208	95	100	100
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	85	100	99
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 Coloboma, ocular, 120433	606608	40	86	73
YARS	Charcot-Marie-Tooth disease, dominant intermediate C, 608323	603623	89	100	100
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	610957	91	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ZAP70	Selective T-cell defect, 269840	176947	113	100	100
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	79	100	100
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069	614064	82	100	100
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	25	97	70
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799	300646	44	100	90
ZEB1	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270	189909	88	100	100
ZEB2	Mowat-Wilson syndrome, 235730	605802	81	100	100
ZFP57	Diabetes mellitus, transient neonatal, 1, 601410	612192	53	99	95
ZFPM2	Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067	603693	78	100	100
ZFYVE26	Spastic paraplegia 15, autosomal recessive, 270700	612012	79	100	97
ZFYVE27	Spastic paraplegia 33, autosomal dominant, 610244	610243	71	100	100
ZIC2	Holoprosencephaly-5, 609637	603073	92	93	88
ZIC3	Heterotaxy, visceral, 1, X-linked 306955 Congenital heart defects, nonsyndromic, 1, X-linked, 306955 VACTERL association, X-linked, 314390	300265	53	100	100
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210	606480	77	100	100
ZMYND10	Ciliary dyskinesia, primary, 22, 615444	607070	60	100	96
ZNF335	?Microcephaly 10, primary, autosomal recessive, 615095	610827	77	100	100
ZNF423	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844	604557	102	100	100
ZNF469	Brittle cornea syndrome 1, 229200	612078	69	100	97
ZNF513	Retinitis pigmentosa 58, 613617	613598	66	100	100
ZNF592	No OMIM phenotype	613624	80	100	99
ZNF644	Myopia 21, autosomal dominant, 614167	614159	67	100	99
ZNF711	Mental retardation, X-linked 97, 300803	314990	39	100	92
ZNF750	Seborrhea-like dermatitis with psoriasiform elements, 610227	610226	77	100	100
ZNF81	Mental retardation, X-linked 45, 300498	314998	36	100	92

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping.
- The statistics above are based on a set of 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x

- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x