Whole Exome Sequencing
Gene package Multiple congenital anomalie, version 4.1, 22-11-2017

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

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
A4GALT	[Blood group, P1Pk system, P(2) phenotype], 111400 [Blood group, P1Pk system, p phenotype], 111400 NOR polyagglutination syndrome, 111400	607922	101	100	100	99
AAAS	Achalasia-addisonianism-alacrimia syndrome, 231550	605378	73	100	100	100
AAGAB	Keratoderma, palmoplantar, punctate type IA, 148600	614888	42	100	98	80
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	59	100	97	86
AARS2	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889	612035	74	100	100	99
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	45	100	95	80
ABAT	GABA-transaminase deficiency, 613163	137150	65	100	98	87
ABCA1	{Coronary artery disease in familial hypercholesterolemia, protection against}, 143890 HDL deficiency, type 2, 604091 Tangier disease, 205400	600046	62	100	97	88
ABCA12	Ichthyosis, congenital 4A, 601277 Ichthyosis, congenital 4B (harlequin), 242500	607800	49	100	98	83
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3, 610921	601615	70	100	98	94

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ABCA4	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 {Macular degeneration, age-related, 2}, 153800 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200	601691	65	100	98	93
ABCB11	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847	603201	46	100	97	83
ABCB4	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803	171060	48	100	98	80
ABCB6	[Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153	605452	64	100	100	97
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	53	100	95	83
ABCC2	Dubin-Johnson syndrome, 237500	601107	53	100	97	84
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850	603234	79	100	100	98
ABCC8	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800	600509	68	100	100	95
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	46	100	95	80
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	56	83	72	70
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	71	100	100	94
ABCG5	Sitosterolemia, 210250	605459	53	100	99	93
ABCG8	{Gallbladder disease 4}, 611465 Sitosterolemia, 210250	605460	93	100	100	98
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	42	100	96	69
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	53	100	99	89
ABL1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib	189980	84	100	100	97
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	97	100	100	100

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ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	64	100	100	98
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	607008	55	100	99	90
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	606885	84	100	100	98
ACADSB	2-methylbutyrylglycinuria, 610006	600301	48	100	95	73
ACADVL	VLCAD deficiency, 201475	609575	81	100	98	93
ACAN	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361	155760	181	100	100	98
ACAT1	Alpha-methylacetoacetic aciduria, 203750	607809	51	100	99	84
ACE	[Angiotensin I-converting enzyme, benign serum increase] {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to} Renal tubular dysgenesis, 267430 {SARS, progression of} {Stroke, hemorrhagic}, 614519	106180	65	100	100	97
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	93	100	100	100
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	71	100	100	94
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944	171640	95	100	100	100
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	86	100	100	96
ACSL4	Mental retardation 63, 300387	300157	51	100	98	87
ACSL6	Myelodysplastic syndrome Myelogenous leukemia, acute	604443	55	100	97	88
ACTA1	Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 ?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3 or recessive, 161800	102610	100	100	100	100
ACTA2	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834	102620	97	100	100	100
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	135	100	100	100
ACTC1	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424	102540	124	100	100	100

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ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	121	100	100	100
ACTN1	Bleeding disorder, platelet-type, 15, 615193	102575	75	100	100	98
ACTN4	Glomerulosclerosis, focal segmental, 1, 603278	604638	64	100	100	98
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	51	100	99	89
ACVR1B	Pancreatic cancer, somatic	601300	55	100	99	90
ACVR2B	Heterotaxy, visceral, 4, autosomal, 613751	602730	74	100	100	96
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2, 600376	601284	74	100	100	100
ACY1	Aminoacylase 1 deficiency, 609924	104620	72	100	100	100
ADA	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700	608958	57	100	98	85
ADA2	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	62	100	99	87
ADAM10	{Alzheimer disease 18, susceptibility to}, 615590 Reticulate acropigmentation of Kitamura, 615537	602192	42	100	94	69
ADAM17	?Inflammatory skin and bowel disease, neonatal, 1, 614328	603639	51	100	96	77
ADAM9	Cone-rod dystrophy 9, 612775	602713	40	100	94	70
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	74	100	100	98
ADAMTS13	Thrombotic thrombocytopenic purpura, familial, 274150	604134	66	97	97	93
ADAMTS17	Weill-Marchesani-like syndrome, 613195	607511	66	97	94	87
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	53	100	98	90
ADAMTS2	Ehlers-Danlos syndrome, type VIIC, 225410	604539	67	100	99	93
ADAMTSL2	Geleophysic dysplasia 1, 231050	612277	52	100	96	78
ADAMTSL4	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, 225100	610113	70	100	99	98
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	67	100	100	98
ADAT3	Mental retardation 36, 615286	615302	84	100	100	100
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	600293	69	98	94	88
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	67	100	100	98
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	60	100	98	90
ADIPOQ	Adiponectin deficiency, 612556	605441	67	100	100	100
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	38	100	90	62

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ADRB2	{Asthma, nocturnal, susceptibility to}, 600807 Beta-2-adrenoreceptor agonist, reduced response to {Obesity, susceptibility to}, 601665	109690	83	100	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	61	100	99	88
AFF2	Mental retardation, FRAXE type, 309548	300806	46	100	96	84
AFG3L2	Spastic ataxia 5, 614487 Spinocerebellar ataxia 28, 610246	604581	54	99	93	79
AGA	Aspartylglucosaminuria, 208400	613228	53	100	98	85
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	49	98	98	91
AGK	Cataract 38, 614691 Sengers syndrome, 212350	610345	44	100	98	83
AGL	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400	610860	53	100	99	89
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	57	100	100	96
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121	603051	48	100	96	74
AGRN	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120	103320	90	99	95	91
AGT	{Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to} Renal tubular dysgenesis, 267430	106150	96	100	100	99
AGTR1	{Hypertension, essential}, 145500 Renal tubular dysgenesis, 267430	106165	48	100	100	98
AGXT	Hyperoxaluria, primary, type 1, 259900	604285	79	100	100	100
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	93	96	96	95
AHI1	Joubert syndrome 3, 608629	608894	51	100	94	73
AICDA	Immunodeficiency with hyper-IgM, type 2, 605258	605257	92	100	100	88
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness 5, 300614	300169	41	100	89	67
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	62	100	100	94
AIP	Pituitary adenoma, ACTH-secreting, 219090 Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634	605555	78	100	100	100
AIPL1	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393	604392	81	100	100	100
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300	607358	55	100	98	83
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	80	100	100	100

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AK2	Reticular dysgenesis, 267500	103020	43	100	100	83
AKAP9	?Long QT syndrome-11, 611820	604001	61	100	99	92
AKR1C2	Obesity, hyperphagia, and developmental delay 46XY sex reversal 8, 614279	600450	65	100	100	96
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555	604741	48	100	92	86
AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500	164730	72	100	100	97
AKT2	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900	164731	70	100	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	49	100	94	75
ALAD	{Lead poisoning, susceptibility to}, 612740 Porphyria, acute hepatic, 612740	125270	71	100	100	97
ALAS2	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, 300752	301300	45	100	97	84
ALB	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999	103600	46	100	97	79
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	54	100	98	88
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	60	100	98	89
ALDH2	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}	100650	79	100	100	100
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	52	100	98	88
ALDH4A1	Hyperprolinemia, type II, 239510	606811	64	100	99	97
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	42	99	94	71
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	603178	71	100	99	93
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	50	100	93	71
ALDOA	Glycogen storage disease XII, 611881	103850	95	100	100	100
ALDOB	Fructose intolerance, 229600	612724	66	100	100	100
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	46	90	76	70
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	59	100	100	99

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ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	108	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	42	100	96	73
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	51	100	100	97
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	63	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	58	100	99	83
ALG8	Congenital disorder of glycosylation, type Ih, 608104	608103	43	100	96	75
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	43	100	96	77
ALMS1	Alstrom syndrome, 203800	606844	82	100	99	96
ALOX12B	Ichthyosis, congenital 2, 242100	603741	79	100	100	100
ALOXE3	Ichthyosis, congenital 3, 606545	607206	82	100	100	98
ALPL	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300	171760	64	100	100	96
ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225	606352	53	100	99	89
ALX1	?Frontonasal dysplasia 3, 613456	601527	57	100	100	99
ALX3	Frontonasal dysplasia 1, 136760	606014	87	100	94	90
ALX4	{Craniosynostosis 5, susceptibility to}, 615529 Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597	605420	83	100	100	100
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	53	100	100	89
AMELX	Amelogenesis imperfecta, type 1E, 301200	300391	106	100	100	100
AMER1	Osteopathia striata with cranial sclerosis, 300373	300647	71	100	100	99
AMH	Persistent Mullerian duct syndrome, type I, 261550	600957	64	100	100	93
AMHR2	Persistent Mullerian duct syndrome, type II, 261550	600956	80	100	100	94
AMN	Megaloblastic anemia-1, Norwegian type, 261100	605799	57	100	99	82
AMPD1	Myopathy due to myoadenylate deaminase deficiency, 615511	102770	48	100	97	85
AMT	Glycine encephalopathy, 605899	238310	73	100	100	98
ANG	Amyotrophic lateral sclerosis 9, 611895	105850	98	100	100	100
ANGPTL3	Hypobetalipoproteinemia, familial, 2, 605019	604774	48	100	99	87
ANK1	Spherocytosis, type 1, 182900	612641	76	100	100	95

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ANK2	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919	106410	53	100	99	93
ANKH	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	605145	59	100	100	98
ANKK1	Dopamine receptor D2, reduced brain density of	608774	70	100	100	99
ANKRD11	KBG syndrome, 148050	611192	89	100	100	100
ANKRD26	Thrombocytopenia 2, 188000	610855	56	100	97	84
ANKS6	Nephronophthisis 16, 615382	615370	56	94	91	82
ANO10	Spinocerebellar ataxia 10, 613728	613726	40	100	92	65
ANO3	Dystonia 24, 615034	610110	43	93	91	77
ANO5	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307	608662	55	100	98	86
ANO6	Scott syndrome, 262890	608663	50	100	98	85
ANOS1	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700	300836	44	100	93	75
ANTXR1	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089	606410	38	98	92	68
ANTXR2	Hyaline fibromatosis syndrome, 228600	608041	46	100	97	76
AP1S1	MEDNIK syndrome, 609313	603531	51	100	100	95
AP1S2	Mental retardation syndromic 5, 304340	300629	44	100	80	65
AP2S1	Hypocalciuric hypercalcemia, type III, 600740	602242	68	100	100	100
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	50	100	95	71
AP4B1	Spastic paraplegia 47, 614066	607245	60	100	100	96
AP4E1	Spastic paraplegia 51, 613744 Stuttering, familial persistent, 1, 184450	607244	50	100	99	87
AP4M1	Spastic paraplegia 50, 612936	602296	75	100	100	98
AP4S1	Spastic paraplegia 52, 614067	607243	37	100	95	69
AP5Z1	Spastic paraplegia 48, 613647	613653	64	100	100	94
APCDD1	Hypotrichosis 1, 605389	607479	76	100	99	91
APOA1	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined Corneal clouding Hypoalphalipoproteinemia, 604091	107680	86	100	100	100
APOA2	Apolipoprotein A-II deficiency {Hypercholesterolemia, familial, modifier of}, 143890	107670	62	100	100	98
APOA5	Hyperchylomicronemia, late-onset, 144650 {Hypertriglyceridemia, susceptibility to}, 145750	606368	119	100	100	100

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APOB	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558	107730	115	100	100	99
APOC2	Hyperlipoproteinemia, type Ib, 207750	608083	62	100	100	100
APOC3	Apolipoprotein C-III deficiency, 614028	107720	81	100	100	100
APRT	Adenine phosphoribosyltransferase deficiency, 614723	102600	52	100	100	100
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	54	100	90	77
AQP2	Diabetes insipidus, nephrogenic, 125800	107777	79	100	100	100
AQP5	Palmoplantar keratoderma, Bothnian type, 600231	600442	66	100	100	100
AR	Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Hypospadias 1, 300633 {Prostate cancer, susceptibility to}, 176807 Spinal and bulbar muscular atrophy of Kennedy, 313200	313700	60	100	95	81
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	55	100	98	85
ARG1	Argininemia, 207800	608313	56	100	100	88
ARHGAP26	Leukemia, juvenile myelomonocytic, somatic, 607785	605370	56	100	100	91
ARHGAP31	Adams-Oliver syndrome 1, 100300	610911	75	100	99	96
ARHGEF10	?Slowed nerve conduction velocity, AD, 608236	608136	72	100	99	91
ARHGEF12	No OMIM phenotype	604763	48	100	97	82
ARHGEF6	Mental retardation 46, 300436	300267	38	100	93	70
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	38	100	95	63
ARID1A	Coffin-Siris syndrome 2, 614607	603024	75	100	98	94
ARID1B	Coffin-Siris syndrome 1, 135900	614556	67	99	97	90
ARL13B	Joubert syndrome 8, 612291	608922	50	100	99	86
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	49	100	100	87
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	32	97	88	57
ARMC4	Ciliary dyskinesia, primary, 23, 615451	615408	50	100	98	84
ARNT	No OMIM phenotype	126110	38	100	95	72
ARSA	Metachromatic leukodystrophy, 250100	607574	105	100	100	100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	611542	49	100	99	90
ARSE	Chondrodysplasia punctata recessive, 302950	300180	63	100	95	84

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ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly 2, 300215 Mental retardation 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	42	89	80	65
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	49	100	97	80
ASB10	Glaucoma 1, open angle, F, 603383	615054	76	100	100	100
ASCC1	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2, 616867	614215	35	100	88	54
ASCL1	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880	100790	120	100	100	100
ASL	Argininosuccinic aciduria, 207900	608310	70	100	100	99
ASNA1	No OMIM phenotype	601913	65	100	100	98
ASNS	Asparagine synthetase deficiency, 615574	108370	59	100	100	92
ASPA	Canavan disease, 271900	608034	41	100	98	81
ASPM	Microcephaly 5, primary, 608716	605481	62	100	100	96
ASPSCR1	Alveolar soft-part sarcoma, 606243	606236	60	100	100	97
ASS1	Citrullinemia, 215700	603470	70	100	100	98
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	77	100	100	97
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	52	99	98	94
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	68	100	96	92
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	45	100	99	84
ATL1	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, 182600	606439	55	100	98	85
ATL3	Neuropathy, hereditary sensory, type IF, 615632	609369	41	100	96	80
ATM	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic	607585	55	100	97	83
ATN1	Dentatorubro-pallidoluysian atrophy, 125370	607462	80	100	100	97
ATP13A2	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, 617225	610513	79	100	100	98

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ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	86	100	100	99
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	91	100	100	100
ATP2A1	Brody myopathy, 601003	108730	76	100	100	98
ATP2A2	Acrokeratosis verruciformis, 101900 Darier disease, 124200	108740	67	100	100	98
ATP2C1	Hailey-Hailey disease, 169600	604384	43	100	96	78
ATP5E	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	606153	56	100	100	100
ATP6V0A2	Cutis laxa, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	56	100	98	87
ATP6V0A4	Renal tubular acidosis, distal, 602722	605239	51	100	96	84
ATP6V1B1	Renal tubular acidosis with deafness, 267300	192132	78	100	100	99
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	44	100	98	84
ATP7B	Wilson disease, 277900	606882	70	100	100	97
ATP8B1	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600	602397	50	100	98	85
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	51	100	100	90
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600	601215	58	100	96	78
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	40	100	91	76
ATXN1	Spinocerebellar ataxia 1, 164400	601556	125	100	100	100
ATXN10	Spinocerebellar ataxia 10, 603516	611150	43	100	97	78
ATXN2	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600 Spinocerebellar ataxia 2, 183090	601517	56	96	92	78
ATXN3	Machado-Joseph disease, 109150	607047	57	100	96	85
ATXN7	Spinocerebellar ataxia 7, 164500	607640	60	99	94	86
ATXN8OS	Spinocerebellar ataxia 8, 608768	603680	No coverage data			
AUH	3-methylglutaconic aciduria, type I, 250950	600529	52	100	100	92
AURKC	Spermatogenic failure 5, 243060	603495	52	100	100	92

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AVP	Diabetes insipidus, neurohypophyseal, 125700	192340	40	95	86	60
AVPR2	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	300538	84	100	100	95
AXIN1	?Caudal duplication anomaly, 607864 Hepatocellular carcinoma, somatic, 114550	603816	82	100	100	98
AXIN2	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615	604025	83	100	100	99
B2M	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600	109700	62	100	100	100
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	42	100	97	76
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	48	79	76	72
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600	606374	73	100	100	100
B3GLCT	Peters-plus syndrome, 261540	610308	47	100	87	64
B4GALNT1	Spastic paraplegia 26, 609195	601873	71	100	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	69	100	100	94
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070	604327	76	100	100	97
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	114	100	100	100
B9D1	Joubert syndrome 27, 617120 ?Meckel syndrome 9, 614209	614144	60	100	100	96
B9D2	Meckel syndrome 10, 614175	611951	55	100	100	99
BAAT	Hypercholanemia, familial, 607748	602938	57	100	98	83
BAG3	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954	603883	95	100	100	100
BANF1	Nestor-Guillermo progeria syndrome, 614008	603811	92	100	100	100
BAP1	Tumor predisposition syndrome, 614327	603089	81	100	100	99
BAX	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065	600040	56	100	99	88
BBS1	Bardet-Biedl syndrome 1, 209900	209901	69	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	55	100	100	98
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	96
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	58	100	99	87
BBS4	Bardet-Biedl syndrome 4, 615982	600374	53	100	96	81
BBS5	Bardet-Biedl syndrome 5, 615983	603650	51	100	99	82
BBS7	Bardet-Biedl syndrome 7, 615984	607590	51	100	98	87
BBS9	Bardet-Biedl syndrome 9, 615986	607968	42	96	94	79

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	48	100	99	83
BCHE	Apnea, postanesthetic	177400	57	100	100	96
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	86	100	100	100
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	49	100	99	86
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923	614901	96	100	100	100
BCL10	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic},, 273300 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}	603517	57	100	100	89
BCL2	Leukemia/lymphoma, B-cell, 2	151430	95	100	100	98
BCL7A	B-cell non-Hodgkin lymphoma, high-grade	601406	50	100	100	93
BCO1	Hypercarotenemia and vitamin A deficiency, 115300	605748	48	100	98	84
BCOR	Microphthalmia, syndromic 2, 300166	300485	77	100	97	92
BCR	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232	151410	84	100	100	98
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	134	100	100	100
BDNF	{Anorexia nervosa, susceptibility to}, 610269 {Bulimia nervosa, age of onset of weight loss in}, 607499 Central hypoventilation syndrome, congenital, 209880 {Memory impairment, susceptibility to} {Obsessive-compulsive disorder, protection against}, 164230	113505	78	100	100	96
BEAN1	Spinocerebellar ataxia 31, 117210	612051	78	100	100	100
BEST1	Bestrophinopathy, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220	607854	63	100	99	96
BFSP1	Cataract 33, multiple types, 611391	603307	69	100	100	94
BFSP2	Cataract 12, multiple types, 611597	603212	61	100	98	87
BICD2	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290	609797	81	100	100	99
BIN1	Myopathy, centronuclear, 255200	601248	65	100	100	97
BLK	Maturity-onset diabetes of the young, type 11, 613375	191305	65	100	100	99

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BLM	Bloom syndrome, 210900	604610	62	100	98	90
BLNK	Agammaglobulinemia 4, 613502	604515	41	100	96	71
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	85	100	100	100
BLOC1S6	Hermansky-pudlak syndrome 9, 614171	604310	36	100	92	61
BLVRA	Hyperbiliverdinemia, 614156	109750	54	100	100	94
BMP1	Osteogenesis imperfecta, type XIII, 614856	112264	69	100	100	99
BMP15	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510	300247	52	100	100	86
BMP2	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200	112261	66	100	99	96
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	72	100	94	84
BMPER	Diaphanospondylodysostosis, 608022	608699	61	100	98	84
BMPR1A	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900	601299	61	100	100	98
BMPR1B	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600	603248	51	100	100	96
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	48	100	99	91
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299	613183	46	100	82	81
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800	613896	56	100	100	98
BRAF	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706	164757	53	100	98	84
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	74	100	100	98
BRIP1	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054	605882	43	100	96	77
BRWD3	Mental retardation 93, 300659	300553	41	100	97	78

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BSCL2	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685	606158	68	100	100	99
BSND	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522	606412	79	100	100	100
BTD	Biotinidase deficiency, 253260	609019	70	100	100	98
BTK	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia 1, 300755	300300	41	100	91	64
BUB1	Colorectal cancer with chromosomal instability, somatic	602452	58	100	98	88
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	51	100	98	86
C12orf57	Temtamy syndrome, 218340	615140	103	100	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, 615035	613541	57	100	100	100
C15orf41	Dyserythropoietic anemia, congenital, type Ib, 615631	615626	42	100	100	86
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	120	100	100	100
C1GALT1C1	Tn polyagglutination syndrome, somatic, 300622	300611	47	100	100	100
C1QA	C1q deficiency, 613652	120550	97	100	100	100
C1QB	C1q deficiency, 613652	120570	85	100	100	96
C1QC	C1q deficiency, 613652	120575	99	100	100	100
C1QTNF5	Retinal degeneration, late-onset, 605670	608752	87	100	100	94
C1S	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174	120580	69	100	100	95
C2	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489	613927	74	100	100	98
C21orf59	Ciliary dyskinesia, primary, 26, 615500	615494	44	100	96	81
C2orf71	Retinitis pigmentosa 54, 613428	613425	91	100	100	100
C3	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378	120700	80	100	100	98
C4A	[Blood group, Rodgers], 614374 C4a deficiency, 614380	120810	228	100	100	99
C4B	C4B deficiency, 614379	120820	215	100	100	100
C4orf26	Amelogenesis imperfecta, type IIA4, 614832	614829	62	100	100	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
C5	C5 deficiency, 609536 [Eculizumab, poor response to], 615749	120900	52	100	97	85
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	54	100	98	88
C6	C6 deficiency, 612446 Combined C6/C7 deficiency	217050	46	100	97	82
C7	C7 deficiency, 610102	217070	57	100	99	90
C8A	C8 deficiency, type I, 613790	120950	51	100	99	89
C8B	C8 deficiency, type II, 613789	120960	69	100	100	95
C8orf37	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500	614477	52	100	100	87
C9	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591	120940	52	100	100	95
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	614260	42	100	97	79
CA12	Hyperchlorhidrosis, isolated, 143860	603263	53	100	100	91
CA2	Osteopetrosis 3, with renal tubular acidosis, 259730	611492	57	100	100	96
CA4	Retinitis pigmentosa 17, 600852	114760	67	100	100	100
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	110	100	100	100
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	51	100	95	81
CABP2	Deafness 93, 614899	607314	51	100	94	84
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	88	100	100	100
CACNA1A	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086	601011	63	100	98	90
CACNA1C	Brugada syndrome 3, 611875 Timothy syndrome, 601005	114205	66	100	99	95
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896	114206	63	100	99	92
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, 300071	300110	63	100	99	91
CACNA1G	Spinocerebellar ataxia 42, 616795	604065	83	100	100	99
CACNA1S	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580	114208	70	100	100	96

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CACNA2D4	Retinal cone dystrophy 4, 610478	608171	57	100	99	93
CACNB2	Brugada syndrome 4, 611876	600003	63	100	100	97
CACNB4	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 Episodic ataxia, type 5, 613855	601949	40	100	97	72
CACNG2	?Mental retardation 10, 614256	602911	78	100	99	94
CALCOCO1	No OMIM phenotype	No id	52	100	100	94
CALM1	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916	114180	48	100	100	93
CALR	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950	109091	97	100	100	100
CALR3	?Cardiomyopathy, hypertrophic, 19, 613875	611414	46	100	97	82
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	90	100	98	94
CANT1	Desbuquois dysplasia 1, 251450	613165	84	100	100	98
CAPN3	Muscular dystrophy, limb-girdle, type 2A, 253600	114240	61	100	99	92
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	73	100	100	93
CARD11	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206	607210	68	100	99	94
CARD14	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723	607211	71	100	100	98
CARD9	Candidiasis, familial, 2, 212050	607212	67	100	100	98
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	42	100	95	69
CASP10	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027	601762	53	100	100	90
CASP8	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 Hepatocellular carcinoma, somatic, 114550 {Lung cancer, protection against}, 211980	601763	65	100	100	90
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938	114251	45	100	97	79

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CASR	{Calcium, serum level of} {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypercalciuric hypercalcemia Hyperparathyroidism, neonatal, 239200 Hypocalcemia, 601198 Hypocalcemia, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980	601199	85	100	100	99
CAT	Acatalasemia, 614097	115500	46	100	97	81
CATSPER1	Spermatogenic failure 7, 612997	606389	88	100	99	96
CAV1	?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 Pulmonary hypertension, primary, 3, 615343	601047	62	100	100	99
CAV3	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Muscular dystrophy, limb-girdle, type IC, 607801 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072	601253	99	100	100	100
CAVIN1	Lipodystrophy, congenital generalized, type 4, 613327	603198	No coverage data			
CBL	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	54	100	100	97
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	70	100	100	96
CBX2	?46XY sex reversal 5, 613080	602770	89	100	98	94
CC2D1A	Mental retardation 3, 608443	610055	79	100	100	99
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	48	100	97	81
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	58	100	98	90
CCDC103	Ciliary dyskinesia, primary, 17, 614679	614677	71	100	100	98
CCDC114	Ciliary dyskinesia, primary, 20, 615067	615038	79	100	100	100
CCDC39	Ciliary dyskinesia, primary, 14, 613807	613798	67	100	97	88
CCDC40	Ciliary dyskinesia, primary, 15, 613808	613799	65	100	100	97
CCDC50	?Deafness 44, 607453	611051	52	100	100	94
CCDC65	Ciliary dyskinesia, primary, 27, 615504	611088	40	100	98	78
CCDC78	Myopathy, centronuclear, 4, 614807	614666	80	100	100	100
CCDC8	3-M syndrome 3, 614205	614145	100	100	100	100

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CCDC88C	Hydrocephalus, nonsyndromic, 236600 ?Spinocerebellar ataxia 40, 616053	611204	67	100	99	94
CCM2	Cerebral cavernous malformations-2, 603284	607929	85	100	96	88
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840	610150	56	100	97	85
CD151	[Blood group, Raph], 179620 Nephropathy with pretibial epidermolysis bullosa and deafness, 609057	602243	66	100	100	100
CD19	Immunodeficiency, common variable, 3, 613493	107265	73	100	100	100
CD247	?Immunodeficiency 25, 610163	186780	50	100	99	85
CD27	Lymphoproliferative syndrome 2, 615122	186711	60	100	100	100
CD2AP	Glomerulosclerosis, focal segmental, 3, 607832	604241	49	100	99	83
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646	606475	65	100	100	96
CD36	{Coronary heart disease, susceptibility to, 7}, 610938 [Macrothrombocytopenia] {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162 Platelet glycoprotein IV deficiency, 608404	173510	48	100	98	82
CD3D	Immunodeficiency 19, 615617	186790	61	100	100	97
CD3E	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615	186830	73	100	100	99
CD3G	Immunodeficiency 17, CD3 gamma deficient, 615607	186740	52	100	100	78
CD4	OKT4 epitope deficiency, 613949	186940	68	100	100	96
CD40	Immunodeficiency with hyper-IgM, type 3, 606843	109535	73	100	100	100
CD40LG	Immunodeficiency, with hyper-IgM, 308230	300386	55	100	100	97
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300	107271	56	100	100	98
CD79A	Agammaglobulinemia 3, 613501	112205	58	100	98	89
CD79B	Agammaglobulinemia 6, 612692	147245	89	100	100	100
CD81	Immunodeficiency, common variable, 6, 613496	186845	83	100	100	97
CD8A	CD8 deficiency, familial, 608957	186910	65	100	98	91
CD96	C syndrome, 211750	606037	52	100	98	88
CDAN1	Dyserythropoietic anemia, congenital, type Ia, 224120	607465	69	100	99	97
CDC6	?Meier-Gorlin syndrome 5, 613805	602627	48	100	100	92
CDC73	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266	607393	56	100	99	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CDH1	{Breast cancer, lobular}, 114480 Endometrial carcinoma, somatic, 608089 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 Ovarian carcinoma, somatic, 167000 {Prostate cancer, susceptibility to}, 176807	192090	60	100	99	95
CDH15	Mental retardation 3, 612580	114019	67	100	99	94
CDH23	Deafness 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067	605516	78	100	100	99
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553	114021	77	100	100	98
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	67	100	100	94
CDK5RAP2	Microcephaly 3, primary, 604804	608201	47	100	95	78
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	46	100	93	78
CDKN1B	Multiple endocrine neoplasia, type IV, 610755	600778	126	100	100	100
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	56	91	83	74
CDKN2A	Melanoma and neural system tumor syndrome, 155755 {Melanoma, cutaneous malignant, 2}, 155601 Orolaryngeal cancer, multiple Pancreatic cancer/melanoma syndrome, 606719	600160	80	100	100	100
CDON	Holoprosencephaly 11, 614226	608707	62	100	100	92
CDSN	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300	602593	89	100	100	100
CDT1	Meier-Gorlin syndrome 4, 613804	605525	66	100	99	92
CEACAM16	Deafness 4B, 614614	614591	61	100	100	98
CEBPA	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626	116897	69	100	87	70
CEBPE	Specific granule deficiency, 245480	600749	79	100	100	100
CEL	Maturity-onset diabetes of the young, type VIII, 609812	114840	88	98	92	88
CENPJ	Microcephaly 6, primary, 608393 ?Seckel syndrome 4, 613676	609279	60	100	100	96
CEP135	Microcephaly 8, primary, 614673	611423	61	100	96	80
CEP152	Microcephaly 9, primary, 614852 Seckel syndrome 5, 613823	613529	52	100	97	84
CEP164	Nephronophthisis 15, 614845	614848	63	100	96	88
CEP19	Morbid obesity and spermatogenic failure, 615703	615586	57	100	100	100

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CEP290	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189	610142	57	100	98	84
CEP41	Joubert syndrome 15, 614464	610523	53	100	99	89
CEP57	Mosaic variegated aneuploidy syndrome 2, 614114	607951	47	100	97	82
CERKL	Retinitis pigmentosa 26, 608380	608381	50	100	98	85
CERS3	Ichthyosis, congenital 9, 615023	615276	46	100	97	82
CES1	Carboxylesterase 1 deficiency	114835	99	100	97	93
CETP	[High density lipoprotein cholesterol level QTL 10], 143470 Hyperalphalipoproteinemia, 143470	118470	61	100	100	99
CFAP53	Heterotaxy, visceral, 6, 614779	614759	73	100	99	94
CFC1	Heterotaxy, visceral, 2, autosomal, 605376	605194	130	100	96	83
CFD	Complement factor D deficiency, 613912	134350	53	100	97	85
CFH	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698	134370	60	100	99	90
CFHR5	Nephropathy due to CFHR5 deficiency, 614809	608593	59	100	100	91
CFI	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439	217030	50	100	97	82
CFL2	Nemaline myopathy 7, 610687	601443	40	100	89	73
CFP	Properdin deficiency, 312060	300383	74	100	89	83
CFTR	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic}, 167800 Sweat chloride elevation without CF	602421	69	100	99	92
CHAT	Myasthenic syndrome, congenital, 6, presynaptic, 254210	118490	77	99	92	86
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	50	100	98	85
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	60	100	99	93
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	65	100	100	100
CHM	Choroideremia, 303100	300390	46	100	91	75
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	56	100	100	96

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CHMP2B	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795	609512	59	100	97	84
CHMP4B	Cataract 31, multiple types, 605387	610897	113	100	100	100
CHN1	Duane retraction syndrome 2, 604356	118423	41	100	96	80
CHRDL1	Megalocornea 1, 309300	300350	36	100	98	77
CHRM3	?Prune belly syndrome, 100100	118494	84	100	100	100
CHRNA1	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930	100690	59	100	100	90
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353	118502	117	100	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	108	100	95	95
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314	100710	81	100	100	98
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375	118507	91	100	100	97
CHRND	Multiple pterygium syndrome, lethal type, 253290 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323	100720	76	100	100	96
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	100	100	100	100
CHRNG	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290	100730	70	100	100	99
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1, 601776	608429	92	100	97	95
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	603799	83	100	100	100
CHST6	Macular corneal dystrophy, 217800	605294	164	100	100	100
CHSY1	Temtamy preaxial brachydactyly syndrome, 605282	608183	61	98	95	93
CHUK	Cocoon syndrome, 613630	600664	45	100	94	70
CIB2	Deafness 48, 609439 Usher syndrome, type IJ, 614869	605564	98	100	100	100
CIITA	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300	600005	76	100	100	97
CISD2	Wolfram syndrome 2, 604928	611507	115	100	100	100
CITED2	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431	602937	117	100	100	100
CLCF1	Cold-induced sweating syndrome 2, 610313	607672	62	100	100	98

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CLCN1	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive	118425	63	100	100	96
CLCN2	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 Leukoencephalopathy with ataxia, 615651	600570	72	100	100	99
CLCN5	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990	300008	52	100	99	91
CLCN7	Osteopetrosis 2, 166600 Osteopetrosis 4, 611490	602727	73	100	100	98
CLCNKA	Bartter syndrome, type 4b, digenic, 613090	602024	101	100	100	98
CLCNKB	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	602023	95	100	100	100
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626	603718	75	100	100	95
CLDN14	Deafness 29, 614035	605608	95	100	100	100
CLDN16	Hypomagnesemia 3, renal, 248250	603959	51	100	99	89
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	75	100	100	100
CLEC7A	{Aspergillosis, susceptibility to}, 614079 Candidiasis, familial, 4, 613108	606264	45	100	98	77
CLIC2	?Mental retardation, syndromic 32, 300886	300138	35	99	89	57
CLMP	Congenital short bowel syndrome, 615237	611693	50	100	100	97
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	76	100	100	94
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	57	100	100	98
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	78	100	98	88
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	81	100	100	100
CLPP	Perrault syndrome 3, 614129	601119	70	100	100	100
CLRN1	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902	606397	54	100	100	87
CNBP	Myotonic dystrophy 2, 602668	116955	37	100	100	78
CNGA1	Retinitis pigmentosa 49, 613756	123825	50	96	90	80
CNGA3	Achromatopsia 2, 216900	600053	84	100	100	98
CNGB1	Retinitis pigmentosa 45, 613767	600724	63	100	99	95

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CNGB3	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200	605080	46	100	97	82
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	106	100	100	95
CNNM4	Jalili syndrome, 217080	607805	90	100	99	96
CNTN1	?Myopathy, congenital, Compton-North, 612540	600016	52	100	99	89
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	48	100	99	89
CNTNAP4	No OMIM phenotype	610518	61	100	98	88
COA5	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500	613920	47	100	100	67
COASY	Neurodegeneration with brain iron accumulation 6, 615643	609855	94	100	100	100
COCH	Deafness 9, 601369	603196	50	98	94	88
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	66	100	99	94
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	53	100	100	89
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	43	100	96	77
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	48	100	96	74
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	56	100	98	85
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	80	100	100	99
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500	120110	68	100	100	98
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	47	100	96	79
COL11A2	Deafness 13, 601868 Deafness 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610	120290	70	100	100	97
COL17A1	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400	113811	65	100	100	95
COL18A1	Knobloch syndrome, type 1, 267750	120328	76	100	100	95

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COL1A1	{Bone mineral density variation QTL, osteoporosis}, 166710 Caffey disease, 114000 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220	120150	87	100	100	98
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710	120160	52	100	98	90
COL2A1	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia	120140	65	100	100	96
COL3A1	Ehlers-Danlos syndrome, type IV, 130050	120180	89	100	100	99
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000	120130	61	100	99	91
COL4A2	{Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 2, 614483	120090	67	100	100	96

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COL4A3	Alport syndrome, 104200 Alport syndrome, 203780 Hematuria, benign familial, 141200	120070	45	98	93	76
COL4A4	Alport syndrome, 203780 Hematuria, familial benign	120131	48	100	97	80
COL4A5	Alport syndrome, 301050	303630	38	99	86	61
COL5A1	Ehlers-Danlos syndrome, classic type, 130000	120215	85	100	98	97
COL5A2	Ehlers-Danlos syndrome, classic type, 130000	120190	48	100	97	81
COL6A1	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090	120220	83	100	100	97
COL6A2	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090	120240	84	100	100	98
COL6A3	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090	120250	74	100	100	96
COL7A1	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705	120120	79	100	100	99
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	57	100	100	91
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	46	100	94	81
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	65	100	100	95
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932	120270	66	100	98	90
COLEC11	3MC syndrome 2, 265050	612502	84	100	100	100
COLQ	Myasthenic syndrome, congenital, 5, 603034	603033	58	100	99	84
COMP	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170	600310	62	100	95	92

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COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	49	100	97	82
COQ6	Coenzyme Q10 deficiency, primary, 6, 614650	614647	60	100	99	88
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	86	100	100	100
COQ8B	Nephrotic syndrome, type 9, 615573	615567	61	100	100	97
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	71	100	100	95
CORIN	Preeclampsia/eclampsia 5, 614595	605236	53	100	100	93
CORO1A	Immunodeficiency 8, 615401	605000	103	100	100	100
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	93	100	100	95
COX14	?Mitochondrial complex IV deficiency, 220110	614478	101	100	100	100
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	43	100	96	76
COX20	Mitochondrial complex IV deficiency, 220110	614698	53	100	100	92
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714	607976	63	100	100	91
COX7B	Linear skin defects with multiple congenital anomalies 2, 300887	300885	55	100	100	98
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	47	100	99	87
CPA6	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418	609562	56	100	96	83
CPN1	Carboxypeptidase N deficiency, 212070	603103	63	100	96	79
CPOX	Coproporphyria, 121300 Harderoporphyria, 121300	612732	46	100	99	85
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	42	100	96	78
CPT1A	CPT deficiency, hepatic, type IA, 255120	600528	65	100	99	93
CPT2	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212	600650	72	100	100	98
CR2	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927	120650	48	100	99	91
CRADD	Mental retardation 34, with variant lissencephaly, 614499	603454	96	100	100	100
CRB1	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105	604210	54	100	100	96

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CRBN	Mental retardation 2, 607417	609262	50	100	96	76
CREB1	Histiocytoma, angiomatoid fibrous, somatic, 612160	123810	53	100	100	96
CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	81	100	99	91
CRELD1	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217	607170	66	100	100	99
CRLF1	Cold-induced sweating syndrome 1, 272430	604237	72	94	90	87
CRTAP	Osteogenesis imperfecta, type VII, 610682	605497	54	100	100	93
CRTC1	Mucoepidermoid salivary gland carcinoma	607536	56	99	91	84
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	93	100	100	100
CRYAA	Cataract 9, multiple types, 604219	123580	61	100	100	97
CRYAB	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869	123590	64	100	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	52	100	100	98
CRYBA4	Cataract 23, 610425	123631	65	100	100	100
CRYBB1	Cataract 17, multiple types, 611544	600929	67	100	100	100
CRYBB2	Cataract 3, multiple types, 601547	123620	83	100	100	100
CRYBB3	Cataract 22, 609741	123630	69	100	100	98
CRYGB	Cataract 39, multiple types, 615188	123670	61	100	100	98
CRYGC	Cataract 2, multiple types, 604307	123680	64	100	100	99
CRYGD	Cataract 4, multiple types, 115700	123690	77	100	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	83	100	100	97
CRYM	Deafness 40, 616357	123740	48	100	99	84
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	164770	63	100	100	97
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4, 300770	306250	39	92	78	62
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5, 614370	138981	73	100	100	99
CSF3R	Neutropenia, severe congenital, 7, 617014	138971	79	100	100	100
CSNK1D	Advanced sleep-phase syndrome, familial, 2, 615224	600864	65	100	100	98
CSPP1	Joubert syndrome 21, 615636	611654	58	100	99	89
CSRP3	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124	600824	70	100	100	100
CST3	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953	604312	62	100	100	85
CSTA	Peeling skin syndrome 4, 607936	184600	45	100	100	88
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	62	100	100	98
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	72	100	100	98

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CTCF	Mental retardation 21, 615502	604167	70	100	100	99
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	78	100	91	86
CTH	Cystathioninuria, 219500 Homocysteine, total plasma, elevated	607657	55	100	99	87
CTHRC1	Barrett esophagus/esophageal adenocarcinoma, 614266	610635	56	100	95	80
CTNNA3	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616	607667	43	100	98	81
CTNNB1	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	47	100	100	92
CTNS	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750	606272	71	100	100	100
CTSA	Galactosialidosis, 256540	613111	83	100	100	98
CTSC	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650	602365	46	100	98	79
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	79	100	100	100
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	70	100	98	93
CTSK	Pycnodysostosis, 265800	601105	45	100	100	93
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	59	100	98	84
CUL3	Pseudohypoaldosteronism, type IIE, 614496	603136	55	100	95	77
CUL4B	Mental retardation, syndromic 15 (Cabezas type), 300354	300304	49	100	93	77
CUL7	3-M syndrome 1, 273750	609577	75	100	100	99
CXCR4	Myelokathexis, isolated WHIM syndrome, 193670	162643	61	83	83	83
CYB5A	?Methemoglobinemia, type IV, 250790	613218	47	100	100	89
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	76	100	100	99
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690	608508	60	99	89	79
CYBB	Chronic granulomatous disease, 306400 Immunodeficiency 34, mycobacteriosis, 300645	300481	39	100	95	79
CYC1	Mitochondrial complex III deficiency, nuclear type 6, 615453	123980	126	100	100	98
CYCS	Thrombocytopenia 4, 612004	123970	48	100	100	100

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CYLD	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606	605018	44	100	96	80
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743	118485	68	100	100	100
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900	610613	122	100	100	100
CYP11B2	Aldosterone to renin ratio raised Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}	124080	122	100	100	100
CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110	609300	80	100	100	98
CYP19A1	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300	107910	49	100	97	79
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300	601771	79	100	100	100
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910	613815	140	100	100	100
CYP24A1	Hypercalcemia, infantile, 1, 143880	126065	69	100	99	87
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	605207	84	100	100	100
CYP26C1	Focal facial dermal dysplasia 4, 614974	608428	74	100	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	82	100	100	100
CYP27B1	Vitamin D-dependent rickets, type I, 264700	609506	89	100	100	99
CYP2A6	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890	122720	136	100	100	100
CYP2B6	{Efavirenz central nervous system toxicity, susceptibility to}, 614546 Efavirenz, poor metabolism of, 614546	123930	84	100	100	100
CYP2C19	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Omeprazole poor metabolizer, 609535 Proguanil poor metabolizer, 609535	124020	77	100	100	98
CYP2C8	Rhabdomyolysis, cerivastatin-induced	601129	59	100	96	84
CYP2C9	Tolbutamide poor metabolizer Warfarin sensitivity, 122700	601130	76	100	98	94
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation, 600081	608713	56	100	100	96
CYP2U1	Spastic paraplegia 56, 615030	610670	51	100	100	91
CYP46A1	No OMIM phenotype	604087	44	100	87	73

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CYP4F22	Ichthyosis, congenital 5, 604777	611495	76	100	100	100
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	60	100	97	89
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, 270800	603711	56	100	100	94
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	82	100	100	100
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538	128239	103	100	100	100
DARS	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281	603084	43	100	93	73
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	45	100	98	81
DBH	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]	609312	72	100	100	99
DBT	Maple syrup urine disease, type II, 248600	248610	77	100	100	97
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	56	100	96	83
DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Mirror movements 1, 157600	120470	48	100	99	89
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	94	100	100	100
DCLRE1C	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450	605988	45	100	96	81
DCN	Corneal dystrophy, congenital stromal, 610048	125255	50	100	100	96
DCTN1	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605	601143	67	100	100	98
DCX	Lissencephaly, 300067 Subcortical laminal heteropia, 300067	300121	47	100	96	83
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740	600811	67	100	100	95
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	55	100	93	82
DDHD1	Spastic paraplegia 28, 609340	614603	67	100	97	85
DDHD2	Spastic paraplegia 54, 615033	615003	57	100	100	93
DDOST	?Congenital disorder of glycosylation, type Ir, 614507	602202	60	100	100	99
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	191311	61	100	100	90
DDX11	Warsaw breakage syndrome, 613398	601150	159	100	100	100
DDX59	Orofaciodigital syndrome V, 174300	615464	50	99	96	83
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	56	100	98	90
DES	Cardiomyopathy, dilated, 1I, 604765 ?Muscular dystrophy, limb-girdle, type 2R, 615325 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400	125660	71	100	100	100

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DFNA5	Deafness 5, 600994	608798	63	100	100	96
DFNB59	Deafness 59, 610220	610219	72	100	100	91
DGKE	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008	601440	78	100	100	91
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions 4, 617070	601465	47	100	92	65
DHCR24	Desmosterolosis, 602398	606418	71	100	100	99
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	84	100	100	100
DHDDS	Retinitis pigmentosa 59, 613861	608172	47	100	100	92
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	65	100	100	96
DHH	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420	605423	79	100	100	100
DHODH	Miller syndrome, 263750	126064	63	100	100	96
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	54	100	99	91
DIABLO	Deafness 64, 614152	605219	68	100	100	96
DIAPH1	Deafness 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	60	100	95	83
DIAPH2	?Premature ovarian failure 2A, 300511	300108	37	100	87	59
DIAPH3	Auditory neuropathy, 1, 609129	614567	50	100	99	85
DICER1	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295	606241	60	100	98	90
DIP2B	Mental retardation, FRA12A type, 136630	611379	46	100	97	81
DIS3L2	Perlman syndrome, 267000	614184	57	100	98	90
DKC1	Dyskeratosis congenita, 305000	300126	45	100	96	83
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	53	100	94	71
DLC1	Colorectal cancer, somatic, 114500	604258	75	100	100	95
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	61	100	100	94
DLG3	Mental retardation 90, 300850	300189	51	100	95	83
DLL3	Spondylocostal dysostosis 1, 277300	602768	63	99	92	85
DLX3	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320	600525	110	100	100	100
DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	38	100	94	71
DMGDH	Dimethylglycine dehydrogenase deficiency, 605850	605849	53	100	99	89

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DMP1	Hypophosphatemic rickets, AR, 241520	600980	60	100	100	96
DMPK	Myotonic dystrophy 1, 160900	605377	69	100	99	93
DNA2	Progressive external ophthalmoplegia with mitochondrial DNA deletions 6, 615156 ?Seckel syndrome 8, 615807	601810	68	100	99	88
DNAAF1	Ciliary dyskinesia, primary, 13, 613193	613190	79	100	99	91
DNAAF2	Ciliary dyskinesia, primary, 10, 612518	612517	89	100	100	100
DNAAF3	Ciliary dyskinesia, primary, 2, 606763	614566	65	100	100	99
DNAAF5	Ciliary dyskinesia, primary, 18, 614874	614864	64	92	84	77
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	603339	54	100	98	87
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	603335	51	100	98	85
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	604366	58	100	98	92
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	605483	75	100	100	98
DNAJB2	Spinal muscular atrophy, distal, 5, 614881	604139	67	100	100	100
DNAJB6	Muscular dystrophy, limb-girdle, type 1E, 603511	611332	57	100	99	91
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	56	100	100	82
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	129	100	100	97
DNAJC6	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528	608375	55	100	99	92
DNAL1	Ciliary dyskinesia, primary, 16, 614017	610062	55	100	100	90
DNASE1L3	Systemic lupus erythematosus 16, 614420	602244	43	100	97	76
DNHD1	No OMIM phenotype	617277	81	100	100	99
DNM1L	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission 1, 614388	603850	53	100	98	85
DNM2	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150	602378	57	100	100	95
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, 604121 Neuropathy, hereditary sensory, type IE, 614116	126375	60	100	98	91
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	71	100	99	92
DOCK6	Adams-Oliver syndrome 2, 614219	614194	72	100	99	98
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	49	100	94	81
DOK7	?Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300	610285	62	95	93	90
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	94	100	100	100
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	54	100	100	95
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	54	93	89	77
DPM2	Congenital disorder of glycosylation, type Iu, 615042	603564	54	100	100	93

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DPM3	Congenital disorder of glycosylation, type Io, 612937	605951	132	100	100	100
DPP6	Mental retardation 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956	126141	49	100	92	78
DPY19L2	Spermatogenic failure 9, 613958	613893	67	100	96	85
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	46	100	99	85
DPYS	Dihydropyrimidinuria, 222748	613326	55	100	99	90
DRC1	Ciliary dyskinesia, primary, 21, 615294	615288	58	100	99	87
DRD2	No OMIM phenotype	126450	80	100	100	100
DRD4	{Attention deficit-hyperactivity disorder}, 143465 Autonomic nervous system dysfunction [Novelty seeking personality], 601696	126452	46	99	83	68
DRD5	{Attention deficit-hyperactivity disorder, susceptibility to}, 143465 {Blepharospasm, primary benign}, 606798 Dystonia, primary cervical	126453	148	100	100	100
DSC2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476	125645	111	100	100	100
DSC3	?Hypotrichosis and recurrent skin vesicles, 613102	600271	57	100	96	79
DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700	125670	65	100	98	87
DSG2	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877	125671	103	100	100	99
DSG4	Hypotrichosis 6, 607903	607892	56	100	100	93
DSP	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655	125647	105	100	100	99
DSPP	Deafness 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500	125485	33	87	61	51
DST	Epidermolysis bullosa simplex 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653	113810	51	100	98	86
DTNA	Left ventricular noncompaction 1, with or without congenital heart defects, 604169	601239	57	100	100	92
DTNBP1	Hermansky-Pudlak syndrome 7, 614076	607145	60	100	97	92
DUOX2	Thyroid dyshormonogenesis 6, 607200	606759	76	100	100	98

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DUOXA2	Thyroid dyshormonogenesis 5, 274900	612772	73	100	100	100
DUSP6	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269	602748	79	100	100	100
DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	49	100	93	68
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	69	100	99	95
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	603297	53	100	97	82
DYRK1A	Mental retardation 7, 614104	600855	54	100	98	87
DYSF	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768	603009	65	100	99	94
DYX1C1	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700	608706	43	100	93	71
EARS2	Combined oxidative phosphorylation deficiency 12, 614924	612799	63	100	100	96
EBP	Chondrodysplasia punctata dominant, 302960 MEND syndrome, 300960	300205	66	100	100	96
ECE1	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500	600423	66	97	97	94
ECEL1	Arthrogryposis, distal, type 5D, 615065	605896	57	100	96	83
ECM1	Urbach-Wiethe disease, 247100	602201	76	100	100	100
EDA	Ectodermal dysplasia 1, hypohidrotic, 305100 Tooth agenesis, selective 1, 313500	300451	59	100	97	86
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, 224900 [Hair morphology 1, hair thickness], 612630	604095	62	100	100	97
EDARADD	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, 614941	606603	52	100	90	82
EDN1	Auriculocondylar syndrome 3, 615706 {High density lipoprotein cholesterol level QTL 7} Question mark ears, isolated, 612798	131240	52	100	98	78
EDN3	Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712 Waardenburg syndrome, type 4B, 613265	131242	69	100	100	100
EDNRA	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300	131243	50	100	97	84

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EDNRB	ABCD syndrome, 600501 {Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580	131244	69	100	100	99
EFEMP1	Doyne honeycomb degeneration of retina, 126600	601548	70	100	100	96
EFEMP2	Cutis laxa, type IB, 614437	604633	76	100	100	100
EFNB1	Craniofrontonasal dysplasia, 304110	300035	55	100	100	97
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536	603892	58	100	99	90
EGF	Hypomagnesemia 4, renal, 611718	131530	53	100	98	84
EGFR	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 ?Inflammatory skin and bowel disease, neonatal, 2, 616069 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980	131550	56	100	100	92
EGLN1	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070	606425	81	100	100	93
EGR2	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 1, 605253	129010	101	100	100	99
EHMT1	Kleefstra syndrome, 610253	607001	81	99	99	98
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	53	100	95	83
EIF2AK4	Pulmonary venoocclusive disease 2, 234810	609280	50	100	97	79
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896	606686	53	100	100	90
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606454	54	100	89	85
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896	606273	40	100	94	79
EIF2B4	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606687	61	100	100	100
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	603945	56	100	100	95
EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305	608546	44	100	98	76
EIF4G1	{Parkinson disease 18}, 614251	600495	73	100	100	99
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731	605367	55	100	99	92
ELANE	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, 202700	130130	67	100	100	98
ELN	Cutis laxa, 123700 Supravalvar aortic stenosis, 185500	130160	70	100	100	98

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ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	56	100	100	95
ELP1	Dysautonomia, familial, 223900	603722	47	100	97	82
EMD	Emery-Dreifuss muscular dystrophy 1, 310300	300384	73	100	100	99
EMG1	Bowen-Conradi syndrome, 211180	611531	57	100	100	100
EMX2	Schizencephaly, 269160	600035	83	100	100	98
ENAM	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650	606585	47	100	100	95
ENG	Telangiectasia, hereditary hemorrhagic, type 1, 187300	131195	76	100	98	94
ENO3	?Glycogen storage disease XIII, 612932	131370	95	100	100	100
ENPP1	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 Hypophosphatemic rickets, 2, 613312 {Obesity, susceptibility to}, 601665	173335	40	96	87	67
ENTPD1	Spastic paraplegia 64, 615683	601752	46	100	97	79
EOGT	Adams-Oliver syndrome 4, 615297	614789	40	100	97	73
EP300	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684	602700	66	100	98	90
EPAS1	Erythrocytosis, familial, 4, 611783	603349	64	100	100	97
EPB41	Elliptocytosis-1, 611804	130500	48	100	99	89
EPB42	Spherocytosis, type 5, 612690	177070	83	100	100	99
EPCAM	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217	185535	51	100	99	87
EPG5	Vici syndrome, 242840	615068	49	100	97	84
EPHA2	Cataract 6, multiple types, 116600	176946	86	100	99	97
EPHB2	{Prostate cancer/brain cancer susceptibility, somatic}, 603688	600997	85	98	98	97
EPHX1	Diphenylhydantoin toxicity ?Fetal hydantoin syndrome Hypercholanemia, familial, 607748 {Preeclampsia, susceptibility to}, 189800	132810	80	100	100	98
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	45	89	85	77
EPX	[Eosinophil peroxidase deficiency], 261500	131399	75	100	100	100
ERBB2	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800 Ovarian cancer, somatic	164870	83	100	99	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ERBB3	Lethal congenital contractural syndrome 2, 607598	190151	74	100	100	98
ERBB4	Amyotrophic lateral sclerosis 19, 615515	600543	46	100	95	75
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	52	100	100	95
ERCC2	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	75	100	100	99
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	133510	56	100	99	89
ERCC4	Fanconi anemia, complementation group Q, 615272 ?XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760	133520	58	100	99	88
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	133530	65	100	99	94
ERCC6	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630	609413	60	100	98	92
ERCC6L2	Bone marrow failure syndrome 2, 615715	615667	44	100	97	82
ERCC8	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621	609412	55	100	93	79
ERF	Chitayat syndrome, 617180 Craniosynostosis 4, 600775	611888	97	100	100	100
ERLIN2	Spastic paraplegia 18, 611225	611605	47	100	99	84
ESCO2	Roberts syndrome, 268300 SC phocomelia syndrome, 269000	609353	48	100	99	87
ESPN	Deafness 36, 609006 Deafness, neurosensory, without vestibular involvement	606351	58	96	82	72
ESR1	{Atherosclerosis, susceptibility to} {Breast cancer}, 114480 Estrogen resistance, 615363 {HDL response to hormone replacement, augmented} {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446	133430	65	100	100	93

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ESRRB	Deafness 35, 608565	602167	85	100	100	99
ETFA	Glutaric acidemia IIA, 231680	608053	41	100	97	78
ETFB	Glutaric acidemia IIB, 231680	130410	68	100	100	100
ETFDH	Glutaric acidemia IIC, 231680	231675	67	100	100	95
ETHE1	Ethylmalonic encephalopathy, 602473	608451	71	100	100	92
ETV6	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216	600618	63	100	99	91
EVC	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	604831	62	96	94	92
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	607261	55	100	97	90
EWSR1	Ewing sarcoma, 612219 Neuroepithelioma, 612219	133450	62	100	97	90
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	59	100	100	87
EXPH5	Epidermolysis bullosa, nonspecific, 615028	612878	52	100	99	95
EXT1	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700	608177	51	100	97	89
EXT2	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	68	100	99	89
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425	605744	100	100	100	98
EYA1	Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780	601653	62	100	99	90
EYA4	Cardiomyopathy, dilated, 1J, 605362 Deafness 10, 601316	603550	44	100	98	80
EYS	Retinitis pigmentosa 25, 602772	612424	56	100	98	91
EZH2	Weaver syndrome, 277590	601573	56	100	99	88
F10	Factor X deficiency, 227600	613872	102	100	99	97
F11	Factor XI deficiency, 612416 Factor XI deficiency, 612416	264900	46	100	99	87
F12	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000	610619	90	100	100	98
F13A1	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050	134570	52	100	100	92
F13B	Factor XIIIB deficiency, 613235	134580	64	100	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
F2	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367 Thrombophilia due to thrombin defect, 188050	176930	76	100	100	98
F5	{Budd-Chiari syndrome}, 600880 Factor V deficiency, 227400 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 Thrombophilia due to activated protein C resistance, 188055 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055	612309	92	100	98	91
F7	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446	613878	82	100	100	99
F8	Hemophilia A, 306700	300841	45	100	99	88
F9	{Deep venous thrombosis, protection against}, 300807 Hemophilia B, 306900 Thrombophilia, due to factor IX defect, 300807 {Warfarin sensitivity}, 122700	300746	53	100	99	83
FA2H	Spastic paraplegia 35, 612319	611026	56	100	100	92
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759	602457	76	100	100	100
FAH	Tyrosinemia, type I, 276700	613871	59	100	100	93
FAM111A	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000	615292	62	100	100	99
FAM111B	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704	615584	52	100	100	97
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	50	100	100	94
FAM161A	Retinitis pigmentosa 28, 606068	613596	45	100	99	87
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690	611062	80	100	100	99
FAM20C	Raine syndrome, 259775	611061	67	100	100	99
FAM58A	STAR syndrome, 300707	300708	50	81	81	80
FAM83H	Amelogenesis imperfecta, type III, 130900	611927	87	100	100	100
FAN1	Interstitial nephritis, karyomegalic, 614817	613534	58	100	100	93
FANCA	Fanconi anemia, complementation group A, 227650	607139	69	100	98	89
FANCB	Fanconi anemia, complementation group B, 300514	300515	46	100	98	85
FANCC	Fanconi anemia, complementation group C, 227645	613899	47	100	94	75
FANCD2	Fanconi anemia, complementation group D2, 227646	613984	53	100	98	87
FANCE	Fanconi anemia, complementation group E, 600901	613976	80	100	100	92
FANCF	Fanconi anemia, complementation group F, 603467	613897	114	100	100	100
FANCG	Fanconi anemia, complementation group G, 614082	602956	88	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FANCI	Fanconi anemia, complementation group I, 609053	611360	52	100	98	87
FANCL	Fanconi anemia, complementation group L, 614083	608111	43	100	94	72
FANCM	No OMIM phenotype	609644	64	100	98	91
FARS2	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, 617046	611592	68	100	100	90
FAS	Autoimmune lymphoproliferative syndrome, type IA, 601859 {Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic	134637	109	100	99	84
FASLG	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980	134638	46	100	100	96
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	69	100	100	98
FBLN1	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4)	135820	69	100	99	96
FBLN5	Cutis laxa 2, 614434 Cutis laxa, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895	604580	69	100	100	94
FBN1	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328	134797	111	100	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	51	100	98	89
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700	611570	82	100	100	100
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	605654	51	100	100	92
FBXO38	Neuronopathy, distal hereditary motor, type IID, 615575	608533	58	100	98	86
FBXO7	Parkinson disease 15, 260300	605648	60	100	100	93
FCGR3A	Immunodeficiency 20, 615707	146740	162	100	100	100
FCGR3B	Neutropenia, alloimmune neonatal	610665	135	100	100	100
FCN3	Immunodeficiency due to ficolin 3 deficiency, 613860	604973	67	100	100	94
FECH	Protoporphyria, erythropoietic, 177000	612386	44	100	98	83
FERMT1	Kindler syndrome, 173650	607900	51	100	97	79
FERMT3	Leukocyte adhesion deficiency, type III, 612840	607901	76	100	99	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FGA	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004	134820	78	100	99	94
FGB	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400	134830	61	100	100	95
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation syndromic 16, 305400	300546	60	100	99	95
FGD4	Charcot-Marie-Tooth disease, type 4H, 609311	611104	47	100	95	79
FGF10	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730	602115	40	100	94	72
FGF14	Spinocerebellar ataxia 27, 609307	601515	47	100	98	83
FGF16	Metacarpal 4-5 fusion, 309630	300827	69	100	100	84
FGF17	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270	603725	71	100	100	100
FGF23	Hypophosphatemic rickets, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900	605380	85	100	100	92
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706	164950	70	100	100	98
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702	600483	94	99	94	94
FGF9	?Multiple synostoses syndrome 3, 612961	600921	60	100	100	100
FGFR1	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440	136350	70	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579	176943	49	100	98	81
FGFR3	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601	134934	84	100	100	98
FGG	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400	134850	55	100	99	92
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	57	98	93	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FHL1	Emery-Dreifuss muscular dystrophy 6, 300696 Myopathy, with postural muscle atrophy, 300696 Reducing body myopathy 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy dominant, 300695	300163	69	100	99	91
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	40	100	96	77
FIGLA	Premature ovarian failure 6, 612310	608697	57	100	100	92
FKBP10	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968	607063	86	100	100	98
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557	614505	69	100	100	91
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	99	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	65	100	100	97
FLCN	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700	607273	72	100	100	100
FLG	{Dermatitis, atopic, susceptibility to, 2}, 605803 Ichthyosis vulgaris, 146700	135940	286	100	100	100
FLNA	Cardiac valvular dysplasia, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	79	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FLNB	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460	603381	71	100	99	94
FLNC	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524	102565	90	100	100	100
FLRT3	Hypogonadotropic hypogonadism 21 with anosmia, 615271	604808	60	100	100	100
FLT3	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626	136351	50	100	98	81
FLT4	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100	136352	81	100	99	97
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	69	100	99	89
FLVCR2	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790	610865	81	100	100	87
FMO3	Trimethylaminuria, 602079	136132	51	100	98	90
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	38	100	94	68
FN1	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101	135600	56	100	98	88
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	85	100	100	100
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482	601090	46	99	90	81
FOXC2	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400	602402	74	100	100	100
FOXE1	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534	602617	80	100	100	97
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	32	82	68	54
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380	601089	83	100	100	100
FOXG1	Rett syndrome, congenital variant, 613454	164874	60	99	91	82
FOXI1	Enlarged vestibular aqueduct, 600791	601093	81	100	100	100

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FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996	605597	86	100	94	88
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705	600838	72	100	100	98
FOXO1	Rhabdomyosarcoma, alveolar, 268220	136533	72	100	100	94
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	56	100	99	89
FOXP2	Speech-language disorder-1, 602081	605317	50	100	98	84
FOXP3	{Diabetes mellitus, type I, susceptibility to}, 222100 Immunodysregulation, polyendocrinopathy, and enteropathy, 304790	300292	59	100	100	91
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	613622	64	100	100	100
FRAS1	Fraser syndrome, 219000	607830	56	100	98	90
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	58	100	99	92
FREM2	Fraser syndrome, 219000	608945	74	100	100	98
FRMD7	Nystagmus 1, congenital, 310700 Nystagmus, infantile periodic alternating, 310700	300628	47	100	94	79
FSCN2	Retinitis pigmentosa 30, 607921	607643	78	100	100	98
FSHB	Hypogonadotropic hypogonadism 24 without anosmia, 229070	136530	58	100	100	100
FSHR	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400	136435	55	100	99	89
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	54	97	93	89
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	90	100	100	100
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460	610966	59	100	100	90
FTSJ1	Mental retardation 9/44, 309549	300499	68	100	100	94
FUCA1	Fucosidosis, 230000	612280	54	100	100	87
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782	137070	68	100	97	96
FUT6	Fucosyltransferase 6 deficiency, 613852	136836	183	100	100	100
FUZ	Neural tube defects, 182940	610622	63	100	100	100
FXN	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300	606829	28	99	62	39
FXYD2	Hypomagnesemia 2, renal, 154020	601814	62	100	100	100

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FYCO1	Cataract 18, 610019	607182	72	100	100	94
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	64	100	100	96
FZD6	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157	603409	50	100	100	95
G6PC	Glycogen storage disease Ia, 232200	613742	64	100	100	95
G6PC3	Dursun syndrome, 612541 Neutropenia, severe congenital 4, 612541	611045	72	100	100	100
G6PD	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162	305900	76	100	100	98
GAA	Glycogen storage disease II, 232300	606800	74	100	100	99
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	52	100	100	90
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	64	100	99	95
GABRG2	{Epilepsy, childhood absence, susceptibility to, 2}, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277	137164	50	92	90	78
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	60	100	100	90
GALC	Krabbe disease, 245200	606890	38	100	94	64
GALE	Galactose epimerase deficiency, 230350	606953	69	100	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	93	100	100	100
GALNS	Mucopolysaccharidosis IVA, 253000	612222	59	100	100	94
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 211900	601756	56	100	100	91
GALT	Galactosemia, 230400	606999	96	100	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	60	100	97	90
GAN	Giant axonal neuropathy-1, 256850	605379	62	100	100	94
GARS	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794	600287	48	100	99	85
GATA1	Anemia, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, 314050 Thrombocytopenia, with or without dyserythropoietic anemia, 300367	305371	54	100	99	94
GATA2	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286	137295	86	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255	131320	75	100	100	100
GATA4	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 ?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429	600576	42	100	86	65
GATA6	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500	601656	61	100	91	74
GATAD1	?Cardiomyopathy, dilated, 2B, 614672	614518	44	100	96	75
GATAD2B	Mental retardation 18, 615074	614998	50	100	100	91
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	44	100	100	89
GBA	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600	606463	145	100	100	100
GBA2	Spastic paraplegia 46, 614409	609471	100	100	100	100
GBE1	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	607839	49	100	95	75
GCDH	Glutaricaciduria, type I, 231670	608801	79	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	45	100	99	87
GCK	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851	138079	73	100	100	100
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446	606857	57	100	99	86
GCM2	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200	603716	68	100	100	97
GCNT2	Adult i phenotype without cataract, 110800 [Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700	600429	62	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GCSH	Glycine encephalopathy, 605899	238330	70	100	91	57
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400	606598	55	100	100	98
GDF1	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854	602880	40	100	90	65
GDF2	Telangiectasia, hereditary hemorrhagic, type 5, 615506	605120	79	100	100	100
GDF3	Klippel-Feil syndrome 3, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	93	100	100	100
GDF5	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 {Osteoarthritis-5}, 612400 Symphalangism, proximal, 1B, 615298	601146	109	100	100	100
GDF6	Klippel-Feil syndrome 1, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094	601147	86	100	100	99
GDI1	Mental retardation 41, 300849	300104	75	100	100	98
GDNF	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300	600837	66	100	100	100
GFAP	Alexander disease, 203450	137780	54	100	100	97
GFER	Myopathy progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	48	100	100	98
GFI1	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, 613107	600871	90	100	100	100
GFI1B	Bleeding disorder, platelet-type, 17, 187900	604383	85	100	100	100
GFM1	Combined oxidative phosphorylation deficiency 1, 609060	606639	52	100	99	88
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, 610542	138292	41	100	97	73

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GGCX	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450	137167	56	100	100	96
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	126	100	100	100
GHR	Growth hormone insensitivity, partial, 604271 {Hypercholesterolemia, familial, modifier of}, 143890 Increased responsiveness to growth hormone Laron dwarfism, 262500	600946	57	100	99	92
GHRHR	Growth hormone deficiency, isolated, type IB, 612781	139191	56	100	99	91
GHSR	Growth hormone deficiency, isolated partial, 615925	601898	93	100	100	100
GIF	Intrinsic factor deficiency, 261000	609342	60	100	100	93
GIGYF2	{Parkinson disease 11}, 607688	612003	47	100	95	79
GIPC3	Deafness 15, 601869	608792	85	98	90	84
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	79	100	100	100
GJA3	Cataract 14, multiple types, 601885	121015	104	100	100	100
GJA5	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770	121013	122	100	100	100
GJA8	Cataract 1, multiple types, 116200	600897	92	100	100	100
GJB1	Charcot-Marie-Tooth neuropathy dominant, 1, 302800	304040	72	100	100	98
GJB2	Bart-Pumphrey syndrome, 149200 Deafness 3A, 601544 Deafness 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500	121011	114	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GJB3	Deafness 2B, 612644 Deafness, with peripheral neuropathy Deafness Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva, 133200	603324	117	100	100	100
GJB4	Erythrokeratodermia variabilis with erythema gyratum repens, 133200	605425	124	100	100	100
GJB6	Deafness 3B, 612643 Deafness 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500	604418	57	100	100	97
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, 613206	608803	52	97	86	75
GK	Glycerol kinase deficiency, 307030	300474	47	100	91	74
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	85	100	100	100
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	67	100	99	87
GLDC	Glycine encephalopathy, 605899	238300	50	100	94	82
GLE1	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310	603371	49	100	99	87
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	81	100	100	99
GLI3	Greig cephalopolysyndactyly syndrome, 175700 {Hypothalamic hamartomas, somatic}, 241800 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700	165240	92	100	100	97
GLIS2	Nephronophthisis 7, 611498	608539	61	100	100	93
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199	610192	85	100	100	98
GLMN	Glomuvenous malformations, 138000	601749	54	100	92	76
GLRA1	Hyperekplexia, hereditary 1 or recessive, 149400	138491	58	100	97	86
GLRB	Hyperekplexia 2, 614619	138492	48	100	100	89
GLRX5	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859	609588	53	100	100	86
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	64	100	93	85

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GLUL	Glutamine deficiency, congenital, 610015	138290	62	100	100	98
GLYCTK	D-glyceric aciduria, 220120	610516	108	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	72	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	69	100	100	100
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352	615320	105	100	100	100
GMPS	No OMIM phenotype	600358	59	100	99	89
GNA11	Hypocalcemia 2, 615361 Hypocalciuric hypercalcemia, type II, 145981	139313	73	100	99	96
GNAI2	Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605	139360	87	100	100	100
GNAI3	Auriculocondylar syndrome 1, 602483	139370	49	100	100	92
GNAL	Dystonia 25, 615073	139312	50	100	98	84
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	55	100	98	84
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	600998	68	100	100	96
GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 Acromegaly, somatic, 102200 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463	139320	86	100	99	97
GNAT1	Night blindness, congenital stationary 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389	139330	107	100	100	100
GNAT2	Achromatopsia 4, 613856	139340	51	100	100	92
GNB4	Charcot-Marie-Tooth disease, dominant intermediate F, 615185	610863	52	100	99	90
GNE	Nonaka myopathy, 605820 Sialuria, 269921	603824	65	100	100	92
GNMT	Glycine N-methyltransferase deficiency, 606664	606628	67	100	100	98
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	59	100	98	90
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	47	100	97	86
GNPTG	Mucolipidosis III gamma, 252605	607838	103	100	98	89
GNRH1	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841	152760	41	100	100	89
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia, 146110	138850	74	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GNS	Mucopolysaccharidosis type IIID, 252940	607664	46	100	98	88
GOLGA5	No OMIM phenotype	606918	43	100	98	83
GORAB	Geroderma osteodysplasticum, 231070	607983	51	100	100	91
GOSR2	Epilepsy, progressive myoclonic 6, 614018	604027	48	100	91	80
GOT1	Aspartate aminotransferase, serum level of, QTL1, 614419	138180	55	100	100	99
GP1BA	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 von Willebrand disease, platelet-type, 177820	606672	77	100	98	96
GP1BB	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200	138720	50	100	97	77
GP6	Bleeding disorder, platelet-type, 11, 614201	605546	68	100	100	97
GP9	Bernard-Soulier syndrome, type C, 231200	173515	87	100	100	100
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	42	100	94	79
GPC6	Omodysplasia 1, 258315	604404	53	100	100	91
GPD1	Hypertriglyceridemia, transient infantile, 614480	138420	57	100	100	98
GPD1L	Brugada syndrome 2, 611777	611778	79	100	100	96
GPHN	Molybdenum cofactor deficiency C, 615501	603930	48	100	99	84
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470	172400	79	100	100	99
GPR143	Nystagmus 6, congenital, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	38	100	89	67
GPR179	Night blindness, congenital stationary (complete), 1E, 614565	614515	91	100	100	100
GPSM2	Chudley-McCullough syndrome, 604213	609245	70	100	99	90
GRHL2	Deafness 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029	608576	56	100	99	87
GRHL3	Van der Woude syndrome 2, 606713	608317	73	100	100	96
GRHPR	Hyperoxaluria, primary, type II, 260000	604296	60	100	100	94
GRIA3	Mental retardation 94, 300699	305915	42	100	97	80
GRIK2	Mental retardation, 6, 611092	138244	63	100	99	92
GRIN1	Mental retardation 8, 614254	138249	75	100	100	97
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	76	100	100	99
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	89	100	100	96
GRIP1	Fraser syndrome, 219000	604597	60	100	99	91
GRK1	Oguchi disease-2, 613411	180381	75	100	100	97
GRM1	Spinocerebellar ataxia 13, 614831	604473	83	100	100	95
GRM6	Night blindness, congenital stationary (complete), 1B, 257270	604096	80	100	92	82

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	101	100	100	100
GRXCR1	Deafness 25, 613285	613283	66	100	100	94
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471	138890	75	100	100	100
GSN	Amyloidosis, Finnish type, 105120	137350	60	100	100	97
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	58	100	100	95
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	41	100	100	85
GTPBP3	Combined oxidative phosphorylation deficiency 23, 616198	608536	85	100	100	99
GUCA1A	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093	600364	66	100	100	100
GUCA1B	Retinitis pigmentosa 48, 613827	602275	96	100	100	100
GUCY1A3	Moyamoya 6 with achalasia, 615750	139396	55	100	98	90
GUCY2C	Diarrhea 6, 614616 Meconium ileus, 614665	601330	55	100	99	84
GUCY2D	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000	600179	73	100	100	99
GUCY2F	No OMIM phenotype	300041	37	100	94	72
GUSB	Mucopolysaccharidosis VII, 253220	611499	74	100	95	85
GYG1	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199	603942	44	100	100	92
GYS1	Glycogen storage disease 0, muscle, 611556	138570	78	100	100	98
GYS2	Glycogen storage disease 0, liver, 240600	138571	46	100	99	88
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data			
H6PD	Cortisone reductase deficiency 1, 604931	138090	96	100	100	100
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	47	100	100	92
HADHA	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015	600890	71	100	100	89
HADHB	Trifunctional protein deficiency, 609015	143450	51	100	98	87
HAMP	Hemochromatosis, type 2B, 613313	606464	72	100	100	100
HARS	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	142810	71	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HARS2	?Perrault syndrome 2, 614926	600783	60	100	100	96
HAX1	Neutropenia, severe congenital 3, 610738	605998	79	100	100	99
HBA1	Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemias, alpha- Thalassemias, alpha-, 604131	141800	178	100	100	100
HBA2	Erythrocytosis Heinz body anemia, 140700 Hemoglobin H disease, nondeletional, 613978 Hypochromic microcytic anemia Thalassemia, alpha-, 604131	141850	134	100	100	100
HBB	Delta-beta thalassemia, 141749 Erythremias, beta- Heinz body anemias, beta-, 140700 Hereditary persistence of fetal hemoglobin, 141749 {Malaria, resistance to}, 611162 Methemoglobinemias, beta- Sickle cell anemia, 603903 Thalassemia-beta, dominant inclusion-body, 603902 Thalassemias, beta-, 613985	141900	87	100	100	100
HBD	Thalassemia due to Hb Lepore Thalassemia, delta-	142000	86	100	100	100
HBG1	Fetal hemoglobin quantitative trait locus 1, 141749	142200	128	100	100	97
HBG2	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749	142250	250	100	100	100
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	35	100	90	54
HCFC1	Mental retardation 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	79	100	100	97
HCN4	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800	605206	89	100	100	98
HCRT	?Narcolepsy 1, 161400	602358	78	100	100	100
HDAC4	No OMIM phenotype	605314	70	100	100	98
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	300272	68	100	99	94
HDAC8	Cornelia de Lange syndrome 5, 300882	300269	33	100	95	65
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	62	100	100	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HERC2	Mental retardation 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	73	100	99	95
HES7	Spondylocostal dysostosis 4, 613686	608059	47	98	89	82
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230	601802	60	100	100	83
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	54	100	99	87
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	69	100	100	94
HFE	{Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200 [Transferrin serum level QTL2], 614193	613609	71	100	100	98
HFE2	Hemochromatosis, type 2A, 602390	608374	76	100	100	100
HFM1	Premature ovarian failure 9, 615724	615684	51	100	95	73
HGD	Alkaptonuria, 203500	607474	45	100	97	77
HGF	Deafness 39, 608265	142409	51	100	99	86
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	48	94	94	85
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	610690	38	100	91	61
HINT1	Neuromyotonia and axonal neuropathy, 137200	601314	64	100	100	100
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	68	100	99	94
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	63	100	99	91
HMBS	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000	609806	62	100	100	99
HMGCL	HMG-CoA lyase deficiency, 246450	613898	57	100	100	94
HMGCS2	HMG-CoA synthase-2 deficiency, 605911	600234	57	100	100	93
HMOX1	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	141250	66	100	100	93
HMX1	Oculoauricular syndrome, 612109	142992	34	93	73	62

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HNF1A	Diabetes mellitus, insulin-dependent, 20, 612520 {Diabetes mellitus, insulin-dependent}, 222100 {Diabetes mellitus, noninsulin-dependent, 2}, 125853 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700	142410	75	100	100	99
HNF1B	Diabetes mellitus, noninsulin-dependent, 125853 {Renal cell carcinoma}, 144700 Renal cysts and diabetes syndrome, 137920	189907	77	100	100	99
HNF4A	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850	600281	72	100	100	98
HNRNPA1	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424	164017	56	100	99	93
HOGA1	Hyperoxaluria, primary, type III, 613616	613597	87	100	100	100
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	104	100	100	100
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432	142958	69	100	100	100
HOXA13	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000	142959	75	86	78	72
HOXB1	Facial paresis, hereditary congenital, 3, 614744	142968	91	100	100	100
HOXC13	Ectodermal dysplasia 9, hair/nail type, 614931	142976	93	100	100	100
HOXD10	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950	142984	79	100	100	100
HOXD13	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713 Syndactyly, type V, 186300 Synpolydactyly 1, 186000	142989	82	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	72	100	100	91
HPGD	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary 1, 259100	601688	55	100	100	87
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	39	100	95	75
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	70	100	100	99
HPS3	Hermansky-Pudlak syndrome 3, 614072	606118	51	100	98	86

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HPS4	Hermansky-Pudlak syndrome 4, 614073	606682	78	100	99	93
HPS5	Hermansky-Pudlak syndrome 5, 614074	607521	50	100	99	85
HPS6	Hermansky-Pudlak syndrome 6, 614075	607522	90	100	100	100
HPSE2	Urofacial syndrome 1, 236730	613469	47	100	95	85
HR	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550	602302	76	100	99	96
HRAS	{Bladder cancer, somatic}, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470	190020	105	100	100	100
HRG	Thrombophilia due to HRG deficiency, 613116 Thrombophilia due to elevated HRG, 613116	142640	59	100	100	88
HSD11B1	Cortisone reductase deficiency 2, 614662	600713	43	100	97	83
HSD11B2	Apparent mineralocorticoid excess, 218030	614232	81	96	90	83
HSD17B10	HDS10 mitochondrial disease, 300438	300256	63	100	100	95
HSD17B3	Pseudohermaphroditism, male, with gynecomastia, 264300	605573	42	100	95	79
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	49	100	97	80
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810	613890	108	100	100	100
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765	607764	90	100	100	100
HSF4	Cataract 5, multiple types, 116800	602438	78	100	100	98
HSPB1	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634	602195	84	100	100	88
HSPB3	?Neuronopathy, distal hereditary motor, type IIC, 613376	604624	96	100	100	100
HSPB8	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590	608014	96	100	97	87
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, 605280	118190	56	100	100	88
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800	142461	78	99	99	99
HTR1A	Periodic fever, menstrual cycle dependent, 614674	109760	107	100	100	100

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HTRA1	CARASIL syndrome, 600142 Cerebral arteriopathy, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149	602194	54	91	82	64
HTRA2	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297	606441	102	100	100	97
HTT	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435	613004	56	99	98	90
HUWE1	Mental retardation syndromic, Turner type, 300706	300697	45	100	93	76
HYAL1	?Mucopolysaccharidosis type IX, 601492	607071	76	100	100	100
HYDIN	Ciliary dyskinesia, primary, 5, 608647	610812	45	100	91	71
HYLS1	Hydrolethalus syndrome, 236680	610693	57	100	100	100
ICK	Endocrine-cerebroosteodysplasia, 612651	612325	49	100	94	79
ICOS	Immunodeficiency, common variable, 1, 607594	604558	42	100	100	88
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	60	100	100	96
IDH3B	Retinitis pigmentosa 46, 612572	604526	74	100	100	100
IDS	Mucopolysaccharidosis II, 309900	300823	63	100	96	85
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	72	100	98	88
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	43	100	86	75
IFITM5	Osteogenesis imperfecta, type V, 610967	614757	92	100	100	100
IFNGR1	{H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948	107470	53	100	98	87
IFT122	Cranioectodermal dysplasia 1, 218330	606045	72	100	100	95
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	70	100	97	91
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	52	100	98	82
IFT43	Cranioectodermal dysplasia 3, 614099	614068	54	100	100	92
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	611177	50	100	93	75
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	59	100	99	84
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	44	100	100	96
IGF1R	Insulin-like growth factor I, resistance to, 270450	147370	75	100	100	95
IGF2R	Hepatocellular carcinoma, somatic, 114550	147280	71	99	97	90

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IGFALS	Acid-labile subunit, deficiency of, 615961	601489	79	100	100	100
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224	602867	42	100	97	82
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320	600502	66	100	100	94
IGLL1	Agammaglobulinemia 2, 613500	146770	96	100	100	100
IGSF1	Hypothyroidism, central, and testicular enlargement, 300888	300137	52	100	99	94
IHH	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500	600726	97	100	100	100
IKBKB	Immunodeficiency 15, 615592	603258	54	100	99	91
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	62	100	100	92
IKZF1	Immunodeficiency, common variable, 13, 616873	603023	78	100	99	94
IKZF5	No OMIM phenotype	606238	71	100	100	97
IL10RA	Inflammatory bowel disease 28, early onset, 613148	146933	73	100	100	96
IL10RB	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, 612567	123889	42	100	98	81
IL11RA	Craniosynostosis and dental anomalies, 614188	600939	69	100	100	99
IL17F	?Candidiasis, familial, 6, 613956	606496	57	100	100	100
IL17RA	Immunodeficiency 51, 613953	605461	80	100	100	100
IL17RD	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267	606807	70	100	100	90
IL1RAPL1	Mental retardation 21/34, 300143	300206	50	100	99	83
IL1RN	{Gastric cancer risk after H. pylori infection}, 137215 Interleukin 1 receptor antagonist deficiency, 612852 {Microvascular complications of diabetes 4}, 612628	147679	55	100	96	83
IL21R	[IgE, elevated level of], 147050 Immunodeficiency, primary, IL21R-related, 615207	605383	78	100	100	95
IL2RA	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367	147730	67	100	100	94
IL2RG	Combined immunodeficiency, moderate, 312863 Severe combined immunodeficiency, 300400	308380	42	100	99	85
IL31RA	Amyloidosis, primary localized cutaneous, 2, 613955	609510	56	100	99	89
IL36RN	Psoriasis 14, pustular, 614204	605507	53	100	100	98
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971	146661	46	100	100	90
ILDR1	Deafness 42, 609646	609739	66	100	98	92

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IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type, 614078	614010	70	100	100	93
IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	67	100	94	87
IMPG2	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581	607056	50	100	98	86
INF2	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237	610982	68	94	92	89
ING1	Squamous cell carcinoma, head and neck, somatic, 275355	601566	92	100	100	100
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	80	100	100	96
INPPL1	Opsismodysplasia, 258480	600829	71	100	99	97
INS	Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176 Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370	176730	82	100	100	100
INSL3	Cryptorchidism, 219050	146738	56	85	78	78
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190	147670	74	100	97	94
INVS	Nephronophthisis 2, infantile, 602088	243305	56	100	98	89
IQCB1	Senior-Loken syndrome 5, 609254	609237	42	100	89	67
IQSEC2	Mental retardation 1/78, 309530	300522	56	99	97	88
IRAK4	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799	606883	53	100	97	77
IRF1	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Nonsmall cell lung cancer, somatic, 211980	147575	71	100	100	94
IRF4	[Skin/hair/eye pigmentation, variation in, 8], 611724	601900	86	100	99	93
IRF6	{Orofacial cleft 6}, 608864 Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300	607199	81	100	100	98
IRF8	Immunodeficiency 32A, mycobacteriosis, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, 614894	601565	87	100	100	92
IRGM	{Inflammatory bowel disease (Crohn disease) 19}, 612278 {Mycobacterium tuberculosis, protection against}, 607948	608212	40	78	78	78
IRX5	Hamamy syndrome, 611174	606195	62	100	100	97

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ISCU	Myopathy with lactic acidosis, hereditary, 255125	611911	56	100	100	90
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	58	100	99	84
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385	606409	43	96	93	76
ITGA2	?Glycoprotein Ia deficiency, 614200	192974	54	100	99	90
ITGA2B	Bleeding disorder, platelet-type, 16, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related	607759	80	100	99	96
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748	605025	80	100	100	98
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730	147556	63	100	99	88
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	600536	68	100	98	94
ITGA8	Renal hypodysplasia/aplasia 1, 191830	604063	43	100	96	74
ITGB2	Leukocyte adhesion deficiency, 116920	600065	77	100	100	99
ITGB3	Bleeding disorder, platelet-type, 16, 187800 Glanzmann thrombasthenia, 273800 {Myocardial infarction, susceptibility to}, 608446 Purpura, posttransfusion Thrombocytopenia, neonatal alloimmune	173470	57	100	99	92
ITGB4	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730	147557	74	99	98	96
ITK	Lymphoproliferative syndrome 1, 613011	186973	53	100	98	79
ITM2B	Dementia, familial British, 176500 Dementia, familial Danish, 117300 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079	603904	49	100	100	89
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	60	100	99	92
IVD	Isovaleric acidemia, 243500	607036	86	100	100	98
IYD	Thyroid dyshormonogenesis 4, 274800	612025	72	100	100	96
JAG1	Alagille syndrome 1, 118450 ?Deafness, congenital heart defects, and posterior embryotoxon Tetralogy of Fallot, 187500	601920	62	100	100	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
JAK2	{Budd-Chiari syndrome, somatic}, 600800 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521	147796	48	100	97	81
JAK3	SCID, T-negative/B-positive type, 600802	600173	64	100	99	96
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	49	100	97	83
JPH2	Cardiomyopathy, hypertrophic, 17, 613873	605267	77	100	100	97
JPH3	?Huntington disease-like 2, 606438	605268	104	100	100	96
JUP	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214	173325	77	100	100	98
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900	607704	73	100	100	95
KANSL1	Koolen-De Vries syndrome, 610443	612452	74	100	100	92
KARS	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness 89, 613916	601421	83	100	99	87
KAT6B	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736	605880	73	100	98	92
KBTBD13	Nemaline myopathy 6, 609273	613727	101	100	100	100
KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	83	100	100	100
KCNA5	Atrial fibrillation, familial, 7, 612240	176267	94	100	100	100
KCNC3	Spinocerebellar ataxia 13, 605259	176264	62	87	64	57
KCND3	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346	605411	114	100	100	97
KCNE1	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695	176261	147	100	100	100
KCNE2	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693	603796	65	100	100	100
KCNE3	Brugada syndrome 6, 613119	604433	73	100	100	100
KCNH2	Long QT syndrome 2, 613688 {Long QT syndrome 2, acquired, susceptibility to}, 613688 Short QT syndrome 1, 609620	152427	82	100	100	97
KCNJ1	Bartter syndrome, type 2, 241200	600359	45	100	100	97
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	133	100	100	100

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KCNJ11	Diabetes mellitus, transient neonatal, 3, 610582 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329	600937	134	100	100	100
KCNJ13	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230	603208	57	100	100	97
KCNJ2	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622	600681	76	100	100	100
KCNJ5	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485	600734	103	100	100	100
KCNK3	Pulmonary hypertension, primary, 4, 615344	603220	95	100	100	97
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292	605874	80	100	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	58	100	98	87
KCNQ1	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Short QT syndrome 2, 609621	607542	77	97	92	90
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data			
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200	602235	81	100	100	97
KCNQ3	Seizures, benign neonatal, type 2, 121201	602232	72	100	98	90
KCNQ4	Deafness 2A, 600101	603537	68	97	92	89
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959	608167	63	100	98	95
KCNV2	Retinal cone dystrophy 3B, 610356	607604	93	100	100	100
KCTD1	Scalp-ear-nipple syndrome, 181270	613420	73	100	98	87
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	102	100	100	99
KDM5C	Mental retardation, syndromic, Claes-Jensen type, 300534	314690	66	100	99	94
KDM6A	Kabuki syndrome 2, 300867	300128	48	100	95	83
KDR	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089	191306	46	100	98	84
KERA	Cornea plana 2, 217300	603288	49	100	100	97
KHDC3L	Hydatidiform mole, recurrent, 2, 614293	611687	86	100	100	100
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	58	100	99	87

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KIF1A	Mental retardation 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, 610357	601255	70	100	100	99
KIF1B	?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700 Pheochromocytoma, 171300	605995	48	100	98	85
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	66	100	99	91
KIF1C	Spastic ataxia 2, 611302	603060	83	100	100	99
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700	608283	51	100	96	80
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546	603213	98	100	100	99
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	59	100	94	75
KIF5A	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, 604187	602821	63	100	99	90
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	66	99	95	89
KIRREL3	Mental retardation 4, 612581	607761	73	100	100	99
KISS1	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842	603286	85	100	99	94
KISS1R	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400	604161	77	100	100	100
KIT	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800	164920	56	100	100	96
KITLG	Deafness 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664	184745	60	100	99	80
KL	{Coronary artery disease, susceptibility to} Tumoral calcinosis, hyperphosphatemic, 211900	604824	75	99	97	96
KLF1	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566	600599	80	100	100	100
KLF11	Maturity-onset diabetes of the young, type VII, 610508	603301	88	100	100	98
KLF6	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807	602053	92	100	100	100

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KLHDC8B	{Hodgkin lymphoma, susceptibility to}, 236000	613169	82	100	100	100
KLHL10	Spermatogenic failure 11, 615081	608778	66	100	100	99
KLHL3	Pseudohypoaldosteronism, type IID, 614495	605775	50	100	94	80
KLHL40	Nemaline myopathy 8, 615348	615340	84	100	100	99
KLHL41	Nemaline myopathy 9, 615731	607701	58	100	100	99
KLHL7	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943	611119	54	100	100	91
KLK4	Amelogenesis imperfecta, type IIA1, 204700	603767	81	100	100	100
KLKB1	Fletcher factor (prekallikrein) deficiency, 612423	229000	53	100	99	87
KLLN	Cowden syndrome 4, 615107	612105	77	100	100	100
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	50	100	99	92
KMT2D	Kabuki syndrome 1, 147920	602113	97	100	100	99
KNL1	Microcephaly 4, primary, 604321	609173	51	100	98	91
KPTN	Mental retardation 41, 615637	615620	73	100	100	94
KRAS	Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	190070	61	100	97	64
KRIT1	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860	604214	51	100	98	81
KRT1	Epidermolytic hyperkeratosis, 113800 Ichthyosis histrix, Curth-Macklin type, 146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962	139350	67	100	100	98
KRT10	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602	148080	72	100	100	99

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KRT12	Meesmann corneal dystrophy, 122100	601687	63	100	100	97
KRT13	White sponge nevus 2, 615785	148065	78	100	98	93
KRT14	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex, recessive 1, 601001 Naegeli-Franceschetti-Jadassohn syndrome, 161000	148066	81	100	100	100
KRT16	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000	148067	95	100	100	100
KRT17	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500	148069	59	100	100	92
KRT18	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600	148070	71	100	100	100
KRT2	Ichthyosis bullosa of Siemens, 146800	600194	72	100	100	97
KRT3	Meesmann corneal dystrophy, 122100	148043	61	100	100	92
KRT4	White sponge nevus 1, 193900	123940	83	100	100	97
KRT5	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex-MP, 131960 Epidermylysis bullosa simplex-MCR, 609352	148040	77	100	100	97
KRT6A	Pachyonychia congenita 3, 615726	148041	138	100	100	100
KRT6B	Pachyonychia congenita 4, 615728	148042	139	100	100	100
KRT6C	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735	612315	116	100	100	100
KRT74	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, 194300	608248	82	100	100	100
KRT8	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600	148060	64	100	98	91
KRT81	Monilethrix, 158000	602153	106	100	100	100
KRT83	?Monilethrix, 158000	602765	89	100	100	100
KRT85	Ectodermal dysplasia 4, hair/nail type, 602032	602767	79	100	100	96
KRT86	Monilethrix, 158000	601928	108	100	100	100
KRT9	Palmoplantar keratoderma, epidermolytic, 144200	607606	69	100	100	100

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L1CAM	CRASH syndrome, 303350 Corpus callosum, partial agenesis of, 304100 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 MASA syndrome, 303350	308840	78	100	100	99
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	47	100	99	89
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855	156225	50	100	98	86
LAMA3	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660	600805	49	100	97	85
LAMA4	Cardiomyopathy, dilated, 1JJ, 615235	600133	50	100	99	86
LAMB1	Lissencephaly 5, 615191	150240	65	100	98	93
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049	150325	97	100	100	100
LAMB3	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650	150310	73	100	100	98
LAMC2	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650	150292	70	100	100	94
LAMC3	Cortical malformations, occipital, 614115	604349	67	100	100	98
LAMP2	Danon disease, 300257	309060	33	100	92	58
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein, 610798	610389	78	100	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	63	100	100	94
LARP7	Alazami syndrome, 615071	612026	50	100	98	85
LARS2	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300	604544	53	100	100	95
LBR	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471	600024	48	100	97	84
LCA5	Leber congenital amaurosis 5, 604537	611408	63	100	100	97
LCAT	Fish-eye disease, 136120 Norum disease, 245900	606967	106	100	100	100
LCT	Lactase deficiency, congenital, 223000	603202	84	100	100	98

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LDB3	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452	605906	66	100	100	97
LDHA	Glycogen storage disease XI, 612933	150000	50	100	96	72
LDHB	[Lactate dehydrogenase-B deficiency], 614128	150100	47	100	100	90
LDLR	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890	606945	117	100	100	100
LDLRAP1	Hypercholesterolemia, familial, 603813	605747	70	100	100	96
LEF1	Sebaceous tumors, somatic	153245	52	100	100	95
LEFTY2	Left-right axis malformations	601877	87	100	100	100
LEMD3	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700	607844	69	100	99	93
LEP	Obesity, morbid, due to leptin deficiency, 614962	164160	73	100	100	100
LEPR	Obesity, morbid, due to leptin receptor deficiency, 614963	601007	61	100	99	91
LFNG	?Spondylocostal dysostosis 3, 609813	602576	63	84	82	79
LGI1	Epilepsy, familial temporal lobe, 1, 600512	604619	64	100	99	92
LHB	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300	152780	81	100	100	100
LHCGR	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410	152790	55	100	98	84
LHFPL5	Deafness 67, 610265	609427	96	100	100	100
LHX3	Pituitary hormone deficiency, combined, 3, 221750	600577	64	100	99	91
LHX4	Pituitary hormone deficiency, combined, 4, 262700	602146	62	100	100	99
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462	607031	56	100	99	88
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	151443	46	100	96	77
LIG1	DNA ligase I deficiency	126391	59	100	100	95
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	62	100	100	100
LIM2	Cataract 19, multiple types, 615277	154045	74	100	100	99
LINS1	Mental retardation 27, 614340	610350	48	100	99	89
LIPA	Cholesteryl ester storage disease, 278000 Wolman disease, 278000	613497	46	100	99	82

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LIPC	{Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797	151670	54	100	98	85
LIPH	Hypotrichosis 7, 604379 Woolly hair 2 with or without hypotrichosis, 604379	607365	40	100	96	78
LIPN	Ichthyosis, congenital 8, 613943	613924	55	100	97	89
LITAF	Charcot-Marie-Tooth disease, type 1C, 601098	603795	65	100	100	100
LMAN1	Combined factor V and VIII deficiency, 227300	601567	47	100	90	74
LMBR1	Acheiropody, 200500 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750 Polydactyly, preaxial type II, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500	605522	38	100	94	64
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380	612625	45	100	92	67
LMF1	Lipase deficiency, combined, 246650	611761	89	100	100	100
LMNA	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210	150330	86	100	100	98
LMNB1	Leukodystrophy, adult-onset, 169500	150340	45	100	98	82
LMX1B	Nail-patella syndrome, 161200	602575	63	100	99	93
LOR	Vohwinkel syndrome with ichthyosis, 604117	152445	64	100	100	100
LOXHD1	Deafness 77, 613079	613072	71	100	99	94
LPAR6	Hypotrichosis 8, 278150 Woolly hair 1, with or without hypotrichosis, 278150	609239	81	100	100	100
LPIN1	Myoglobinuria, acute recurrent, 268200	605518	53	100	98	84
LPIN2	Majeed syndrome, 609628	605519	53	100	99	88

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LPL	Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11] Lipoprotein lipase deficiency, 238600	609708	74	100	100	95
LPP	Leukemia, acute myeloid, 601626 Lipoma	600700	61	100	100	98
LRAT	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341	604863	105	100	100	100
LRBA	Immunodeficiency, common variable, 8, with autoimmunity, 614700	606453	47	100	97	82
LRIG2	Urofacial syndrome 2, 615112	608869	56	100	99	93
LRIT3	Night blindness, congenital stationary (complete), 1F, 615058	615004	61	100	100	98
LRMDA	Albinism, oculocutaneous, type VII, 615179	614537	56	100	94	83
LRP2	Donnai-Barrow syndrome, 222448	600073	47	100	97	84
LRP4	Cenani-Lenz syndactyly syndrome, 212780 ?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305	604270	68	100	99	95
LRP5	[Bone mineral density variability 1], 601884 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 {Osteoporosis}, 166710 Osteosclerosis, 144750 van Buchem disease, type 2, 607636	603506	81	100	98	96
LRPAP1	Myopia 23, 615431	104225	76	100	100	99
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	44	100	95	74
LRRC6	Ciliary dyskinesia, primary, 19, 614935	614930	58	100	98	83
LRRC8A	?Agammaglobulinemia 5, 613506	608360	111	100	100	100
LRRK2	{Parkinson disease 8}, 607060	609007	61	100	98	88
LRSAM1	Charcot-Marie-Toothe disease, axonal, type 2P, 614436	610933	56	100	96	87
LRTOMT	Deafness 63, 611451	612414	60	100	100	98
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	70	100	100	99
LTBP3	Dental anomalies and short stature, 601216	602090	86	100	99	97
LTBP4	Cutis laxa, type IC, 613177	604710	80	100	99	98
LYST	Chediak-Higashi syndrome, 214500	606897	55	100	99	91

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LYZ	Amyloidosis, renal, 105200	153450	47	100	100	93
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	57	100	98	83
LZTS1	Esophageal squamous cell carcinoma, 133239	606551	90	100	100	100
MAD1L1	Lymphoma, somatic Prostate cancer, somatic, 176807	602686	69	100	100	98
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	52	85	80	75
MAFB	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300	608968	133	100	100	100
MAGEL2	Schaaf-Yang syndrome, 615547	605283	107	100	100	100
MAGT1	Immunodeficiency, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	42	100	99	79
MAK	Retinitis pigmentosa 62, 614181	154235	67	100	99	92
MAML2	Mucoepidermoid salivary gland carcinoma	607537	69	100	100	98
MAMLD1	Hypospadias 2, 300758	300120	65	100	97	93
MAN1B1	Mental retardation 15, 614202	604346	90	100	100	98
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	75	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	59	100	98	87
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	41	100	96	80
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	55	100	100	91
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	69	100	92	86
MAP3K1	46XY sex reversal 6, 613762	600982	53	100	99	90
MAP3K8	Lung cancer, somatic, 211980	191195	51	100	100	97
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 {Parkinson disease, susceptibility to}, 168600 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540	157140	63	100	100	94
MARS2	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, 611390	609728	107	100	100	100
MARVELD2	Deafness 49, 610153	610572	70	100	100	97
MASP1	3MC syndrome 1, 257920	600521	70	100	100	92
MASP2	MASP2 deficiency, 613791	605102	60	100	98	91
MASTL	No OMIM phenotype	608221	49	99	98	88
MAT1A	Hypermethioninemia, persistent, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, 250850	610550	65	100	100	99

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MATN3	Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600 Spondyloepimetaphyseal dysplasia, 608728	602109	51	98	87	71
MATR3	Amyotrophic lateral sclerosis 21, 606070	164015	58	100	100	95
MBD5	Mental retardation 1, 156200	611472	58	100	100	97
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, 308800 ?Olmsted syndrome, 300918	300294	42	100	98	81
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200	607397	90	100	100	100
MC4R	Obesity, 601665	155541	56	100	100	100
MCC	Colorectal cancer, somatic, 114500	159350	61	100	99	90
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	48	100	99	83
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	46	100	99	86
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120	608419	55	100	100	80
MCFD2	Factor V and factor VIII, combined deficiency of, 613625	607788	63	100	100	84
MCM4	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981	602638	56	100	99	91
MCM6	Lactase persistence/nonpersistence, 223100	601806	48	100	98	81
MCOLN1	Mucolipidosis IV, 252650	605248	84	100	100	100
MCPH1	Microcephaly 1, primary, 251200	607117	59	94	94	85
MECP2	{Autism susceptibility 3}, 300496 Encephalopathy, neonatal severe, 300673 Mental retardation syndromic, Lubs type, 300260 Mental retardation, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	88	100	100	94
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, 300895 Opitz-Kaveggia syndrome, 305450	300188	57	100	100	97
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808	608771	56	100	97	87
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	69	100	100	94
MED23	Mental retardation 18, 614249	605042	47	100	98	84
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589	610197	75	100	100	100
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4) Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	67	100	99	96

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MEFV	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100	608107	78	100	100	99
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399	612453	61	100	99	90
MEGF8	Carpenter syndrome 2, 614976	604267	80	100	100	99
MEN1	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic	613733	111	100	100	100
MEOX1	Klippel-Feil syndrome 2, 214300	600147	66	100	100	98
MERTK	Retinitis pigmentosa 38, 613862	604705	64	100	99	93
MESP2	Spondylocostal dysostosis 2, 608681	605195	80	100	100	98
MET	?Deafness 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 {Osteofibrous dysplasia, susceptibility to}, 607278 Renal cell carcinoma, papillary, 1, familial and somatic, 605074	164860	47	100	98	88
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152	608507	74	100	100	95
MFRP	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	606227	76	100	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	50	100	98	81
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	80	100	100	100
MGME1	Mitochondrial DNA depletion syndrome 11, 615084	615076	44	94	93	81
MGP	Keutel syndrome, 245150	154870	47	100	94	82
MIB1	Left ventricular noncompaction 7, 615092	608677	49	100	98	79
MICU1	Myopathy with extrapyramidal signs, 615673	605084	36	100	94	58
MID1	Opitz GBBB syndrome, type I, 300000	300552	70	100	98	85
MINPP1	Thyroid carcinoma, follicular, 188470	605391	88	100	100	100
MIP	Cataract 15, multiple types, 615274	154050	85	100	100	100
MIR17HG	Feingold syndrome 2, 614326	609415	No coverage data			
MIR184	EDICT syndrome, 614303	613146	No coverage data			
MIR96	Deafness 50, 613074	611606	No coverage data			

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MITF	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470	156845	58	100	100	90
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	59	100	100	99
MKL1	Megakaryoblastic leukemia, acute	606078	72	98	95	93
MKRN3	Precocious puberty, central, 2, 615346	603856	86	100	100	100
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	81	100	100	97
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	49	100	98	86
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320	120436	122	100	100	100
MLH3	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089	604395	50	100	100	95
MLLT10	Leukemia, acute myeloid, 601626	602409	55	100	98	92
MLLT11	No OMIM phenotype	604684	55	100	100	100
MLPH	Griscelli syndrome, type 3, 609227	606526	69	100	100	97
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	56	100	97	87
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	59	100	100	97
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110	607568	50	100	99	84
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	76	100	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	51	100	98	78
MMP1	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, modifier of}, 226600	120353	45	100	100	92
MMP13	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111	600108	58	100	99	90
MMP2	Multicentric osteolysis, nodulosis, and arthropathy, 259600	120360	70	100	100	95
MMP20	Amelogenesis imperfecta, type IIA2, 612529	604629	46	100	99	84
MMP21	Heterotaxy, visceral, 7, autosomal, 616749	608416	52	100	94	83

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MMP9	Metaphyseal anadysplasia 2, 613073	120361	71	100	100	99
MN1	Meningioma, 607174	156100	109	100	100	100
MNX1	Currarino syndrome, 176450	142994	37	80	72	66
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	69	100	100	98
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	50	100	100	90
MOG	?Narcolepsy 7, 614250	159465	68	100	100	93
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	88	100	100	99
MPC1	Mitochondrial pyruvate carrier deficiency, 614741	614738	43	100	93	64
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	60	100	100	97
MPDZ	Hydrocephalus, nonsyndromic 2, 615219	603785	49	100	97	85
MPI	Congenital disorder of glycosylation, type Ib, 602579	154550	103	100	100	99
MPL	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498	159530	64	100	100	98
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050	609188	48	100	100	97
MPO	{Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers} Myeloperoxidase deficiency, 254600	606989	92	100	100	100
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	137960	65	100	100	99
MPZ	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 605253 Roussy-Levy syndrome, 180800	159440	75	100	100	99
MRAP	Glucocorticoid deficiency 2, 607398	609196	75	100	100	100
MRE11	Ataxia-telangiectasia-like disorder, 604391	600814	43	100	91	72
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582	607118	44	100	97	78
MRPS16	Combined oxidative phosphorylation deficiency 2, 610498	609204	58	100	100	98
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	61	100	99	84
MS4A1	Immunodeficiency, common variable, 5, 613495	112210	63	100	99	88
MSH2	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320	609309	112	100	100	100
MSH3	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4, 617100	600887	55	100	98	87

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MSH6	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300	600678	148	100	100	100
MSR1	Barrett esophagus/esophageal adenocarcinoma, 614266 Prostate cancer, hereditary, 176807	153622	63	100	94	86
MSRB3	Deafness 74, 613718	613719	47	100	99	81
MSTN	Muscle hypertrophy, 614160	601788	65	100	99	91
MSX1	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600	142983	80	100	100	100
MSX2	Craniosynostosis 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550	123101	68	100	100	100
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250	156540	51	100	97	78
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947	611766	52	100	96	72
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	65	100	100	98
MTM1	Myotubular myopathy, 310400	300415	38	100	95	67
MTMR2	Charcot-Marie-Tooth disease, type 4B1, 601382	603557	51	100	96	81
MTO1	Combined oxidative phosphorylation deficiency 10, 614702	614667	82	96	94	92
MTPAP	?Spastic ataxia 4, 613672	613669	68	100	99	91
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	52	100	98	85
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	61	100	98	92
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	49	100	99	89
MUC1	Medullary cystic kidney disease 1, 174000	158340	80	100	100	94
MUSK	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325	601296	58	100	99	92
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	56	100	98	86
MUTYH	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, with pilomatricomas, 132600 Gastric cancer, somatic, 613659	604933	114	100	100	100

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MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	58	100	100	91
MXI1	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807	600020	37	94	83	61
MYBPC1	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915	160794	43	100	96	74
MYBPC3	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396	600958	98	100	100	99
MYCN	Feingold syndrome 1, 164280	164840	79	100	97	93
MYD88	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260	602170	94	100	100	100
MYF6	Myopathy, centronuclear, 3, 614408	159991	76	100	100	100
MYH11	Aortic aneurysm, familial thoracic 4, 132900	160745	76	100	99	91
MYH14	Deafness 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369	608568	72	100	99	95
MYH2	Proximal myopathy and ophthalmoplegia, 605637	160740	61	100	100	95
MYH3	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110	160720	64	100	100	95
MYH6	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090	160710	97	99	98	98
MYH7	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, 608358 Myopathy, myosin storage, 255160 Scapuloperoneal syndrome, myopathic type, 181430	160760	114	100	100	100
MYH7B	No OMIM phenotype	609928	79	100	100	99
MYH8	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300	160741	58	100	100	94

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MYH9	Deafness 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249	160775	66	100	98	94
MYL2	Cardiomyopathy, hypertrophic, 10, 608758	160781	103	100	100	100
MYL3	Cardiomyopathy, hypertrophic, 8, 608751	160790	67	100	100	100
MYLK	Aortic aneurysm, familial thoracic 7, 613780	600922	79	100	100	96
MYLK2	Cardiomyopathy, hypertrophic, 1, digenic, 192600	606566	69	100	100	99
MYO15A	Deafness 3, 600316	602666	74	100	98	92
MYO1A	No OMIM phenotype	601478	64	100	100	99
MYO1E	Glomerulosclerosis, focal segmental, 6, 614131	601479	52	100	98	87
MYO3A	Deafness 30, 607101	606808	55	100	98	85
MYO5A	Griscelli syndrome, type 1, 214450	160777	51	100	97	84
MYO5B	Microvillus inclusion disease, 251850	606540	68	100	99	90
MYO6	Deafness 22, 606346 Deafness 22, with hypertrophic cardiomyopathy, 606346 Deafness 37, 607821	600970	63	100	98	84
MYO7A	Deafness 11, 601317 Deafness 2, 600060 Usher syndrome, type 1B, 276900	276903	68	100	98	95
MYOC	Glaucoma 1A, primary open angle, 137750	601652	105	100	100	96
MYOT	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920	604103	54	100	99	87
MYOZ2	Cardiomyopathy, hypertrophic, 16, 613838	605602	51	100	100	93
MYPN	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, 617336	608517	51	100	98	88
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	56	100	95	92
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	72	100	100	100
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	80	100	97	90

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NAGS	N-acetylglutamate synthase deficiency, 237310	608300	57	100	100	99
NALCN	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	611549	50	100	99	88
NANOS1	Spermatogenic failure 12, 615413	608226	52	98	93	82
NBAS	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800	608025	47	100	97	82
NBEAL2	Gray platelet syndrome, 139090	614169	90	100	99	99
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	52	100	95	76
NCF1	Chronic granulomatous disease due to deficiency of NCF-1, 233700	608512	98	100	100	98
NCF2	Chronic granulomatous disease due to deficiency of NCF-2, 233710	608515	63	100	100	90
NCF4	?Granulomatous disease, chronic, cytochrome b-positive, type III, 613960	601488	67	100	100	99
NCOA4	No OMIM phenotype	601984	61	100	100	96
NCSTN	Acne inversa, familial, 1, 142690	605254	63	100	100	93
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	61	100	99	92
NDN	Prader-Willi syndrome, 176270	602117	93	100	100	100
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	57	100	100	100
NDRG1	Charcot-Marie-Tooth disease, type 4D, 601455	605262	65	100	99	91
NDUFA1	Mitochondrial complex I deficiency, 252010 dominant	300078	83	100	100	100
NDUFA10	Leigh syndrome, 256000	603835	56	98	89	65
NDUFA11	Mitochondrial complex I deficiency, 252010 dominant	612638	72	100	100	100
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000	614530	48	100	100	79
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	87	100	100	100
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency, 256000	603834	62	100	100	96
NDUFAF1	Mitochondrial complex I deficiency, 252010 dominant	606934	49	100	99	86
NDUFAF2	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	609653	63	100	90	77
NDUFAF3	Mitochondrial complex I deficiency, 252010 dominant	612911	79	100	100	100
NDUFAF4	Mitochondrial complex I deficiency, 252010 dominant	611776	72	100	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010 dominant	612360	54	100	100	89
NDUFAF6	Leigh syndrome due to mitochondrial complex I deficiency, 256000	612392	39	100	97	73
NDUFB3	Mitochondrial complex I deficiency, 252010 dominant	603839	28	100	81	47
NDUFS1	Mitochondrial complex I deficiency, 252010 dominant	157655	49	100	98	77
NDUFS2	Mitochondrial complex I deficiency, 252010 dominant	602985	59	100	100	97

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NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	603846	73	100	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	602694	56	100	100	90
NDUFS6	Mitochondrial complex I deficiency, 252010 dominant	603848	52	100	100	96
NDUFS7	Leigh syndrome, 256000	601825	64	100	100	97
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	91	100	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010 dominant	161015	95	100	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010 dominant	600532	47	100	96	78
NEB	Nemaline myopathy 2, 256030	161650	73	100	98	88
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	600644	74	100	100	100
NECTIN4	Ectodermal dysplasia-syndactyly syndrome 1, 613573	609607	76	100	100	98
NEFL	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684	162280	106	100	100	100
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604588	45	100	95	75
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	99	100	100	100
NEUROD1	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394	601724	102	100	100	100
NEUROG3	Diarrhea 4, malabsorptive, congenital, 610370	604882	121	100	100	100
NEXMIF	Mental retardation 98, 300912	300524	51	100	100	98
NEXN	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876	613121	74	100	100	96
NF1	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	41	98	87	67
NF2	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, 162091	607379	84	100	100	100
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	78	100	99	90
NFKB2	Immunodeficiency, common variable, 10, 615577	164012	102	100	100	99
NFKBIA	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132	164008	84	100	100	98
NFU1	Multiple mitochondrial dysfunctions syndrome 1, 605711	608100	48	100	100	88
NGF	Neuropathy, hereditary sensory and autonomic, type V, 608654	162030	104	100	100	100

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NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291	611290	40	100	99	80
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	93	100	100	100
NHP2	Dyskeratosis congenita 2, 613987	606470	66	100	100	100
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	44	100	92	84
NIN	?Seckel syndrome 7, 614851	608684	56	100	98	84
NIPA1	Spastic paraplegia 6, 600363	608145	65	100	96	85
NIPAL4	Ichthyosis, congenital 6, 612281	609383	66	100	100	98
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	50	100	96	80
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550	600635	54	100	100	96
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432	600584	84	100	100	100
NKX2-6	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095	611770	87	100	100	100
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	602183	77	100	100	100
NLGN4X	{Asperger syndrome susceptibility 2}, 300497 {Autism susceptibility 2}, 300495 Mental retardation, 300495	300427	153	100	100	100
NLRP12	Familial cold autoinflammatory syndrome 2, 611762	609648	85	100	100	98
NLRP3	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900	606416	85	100	100	100
NLRP7	Hydatidiform mole, recurrent, 1, 231090	609661	109	100	100	99
NME8	Ciliary dyskinesia, primary, 6, 610852	607421	50	100	98	83
NMNAT1	Leber congenital amaurosis 9, 608553	608700	62	100	100	94
NNT	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736	607878	48	100	98	84
NOBOX	Premature ovarian failure 5, 611548	610934	75	100	100	100
NOD2	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321	605956	73	100	99	94

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NODAL	Heterotaxy, visceral, 5, 270100	601265	79	100	100	100
NOG	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumb and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570	602991	132	100	100	100
NOL3	Myoclonus, familial cortical, 614937	605235	92	100	100	100
NOP10	Dyskeratosis congenita 1, 224230	606471	121	100	100	100
NOP56	Spinocerebellar ataxia 36, 614153	614154	75	100	100	100
NOTCH1	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730	190198	87	100	99	98
NOTCH2	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500	600275	75	100	100	96
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293	600276	75	99	96	92
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	58	100	100	93
NPC2	Niemann-pick disease, type C2, 607625	601015	64	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	39	100	92	67
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	47	100	97	82
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	67	100	100	98
NPHS1	Nephrotic syndrome, type 1, 256300	602716	70	100	100	98
NPHS2	Nephrotic syndrome, type 2, 600995	604766	59	100	100	96
NPM1	Leukemia, acute myeloid, somatic, 601626	164040	64	100	100	91
NPPA	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745	108780	88	100	100	100
NPR2	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255	108961	83	100	100	100
NR0B1	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018	300473	77	100	100	99
NR0B2	Obesity, mild, early-onset, 601665	604630	62	100	100	100

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NR2E3	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131	604485	75	100	100	100
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	82	100	98	91
NR3C1	Glucocorticoid resistance, 615962	138040	62	100	99	91
NR3C2	Hypertension, early-onset, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, 177735	600983	68	100	99	95
NR4A3	Chondrosarcoma, extraskeletal myxoid, 612237	600542	68	100	100	97
NR5A1	Adrenocortical insufficiency, 612964 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957 46, XX sex reversal 4, 617480 46XY sex reversal 3, 612965	184757	72	100	98	95
NRAS	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470	164790	42	100	100	91
NRL	Retinal degeneration, clumped pigment type Retinitis pigmentosa 27, 613750	162080	71	100	100	100
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	67	100	99	93
NSD1	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	56	100	99	92
NSD3	Leukemia, acute myeloid, 601626	607083	41	100	94	73
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	55	100	96	80
NSMF	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838	608137	73	100	95	95
NSUN2	Mental retardation 5, 611091	610916	58	100	94	83
NT5C2	Spastic paraplegia 45, 613162	600417	42	100	95	75
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency, 266120	606224	46	100	96	81
NT5E	Calcification of joints and arteries, 211800	129190	56	100	100	92
NTF4	Glaucoma 1, open angle, 1O, 613100	162662	85	100	100	100
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	70	100	100	98

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NTRK2	Obesity, hyperphagia, and developmental delay, 613886	600456	61	100	97	86
NUBPL	Mitochondrial complex I deficiency, 252010 dominant	613621	53	100	100	89
NUMA1	Leukemia, acute promyelocytic, somatic, 612376	164009	81	100	98	93
NUP214	Leukemia, T-cell acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, somatic, 601626	114350	68	100	99	89
NUP62	Striatonigral degeneration, infantile, 271930	605815	89	100	100	100
NYX	Night blindness, congenital stationary (complete), 1A, 310500	300278	72	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	53	100	100	89
OBSL1	3-M syndrome 2, 612921	610991	76	100	100	98
OCA2	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	611409	60	100	99	89
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	65	100	100	97
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	36	100	92	67
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	42	100	97	81
OGG1	Renal cell carcinoma, clear cell, somatic, 144700	601982	68	100	100	100
OPA1	Behr syndrome, 210000 {Glaucoma, normal tension, susceptibility to}, 606657 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250	605290	46	100	96	77
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	66	100	100	100
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	44	100	92	71
OPLAH	5-oxoprolinase deficiency, 260005	614243	77	100	99	97
OPN1LW	Blue cone monochromacy, 303700 Colorblindness, protan, 303900	300822	93	100	100	100
OPN1MW	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800	300821	103	100	100	99
OPN1SW	Colorblindness, tritan, 190900	613522	57	100	100	100
OPTN	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657	602432	56	100	98	87

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ORAI1	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883	610277	88	99	99	95
ORC1	Meier-Gorlin syndrome 1, 224690	601902	59	100	98	87
ORC4	Meier-Gorlin syndrome 2, 613800	603056	51	100	97	74
ORC6	Meier-Gorlin syndrome 3, 613803	607213	48	100	100	82
OSMR	Amyloidosis, primary localized cutaneous, 1, 105250	601743	54	100	99	88
OSTM1	Osteopetrosis 5, 259720	607649	58	100	100	88
OTC	Ornithine transcarbamylase deficiency, 311250	300461	46	100	98	78
OTOA	Deafness 22, 607039	607038	63	100	99	92
OTOF	Auditory neuropathy, 1, 601071 Deafness 9, 601071	603681	71	100	100	99
OTOG	Deafness 18B, 614945	604487	73	100	100	98
OTOGL	Deafness 84B, 614944	614925	54	100	96	81
OTX2	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125	600037	80	100	100	100
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	601424	50	100	99	83
P2RX1	Bleeding disorder due to P2RX1 defect, somatic, 609821	600845	71	100	100	100
P2RX2	Deafness 41, 608224	600844	77	100	100	99
P2RY12	Bleeding disorder, platelet-type, 8, 609821	600515	61	100	100	100
P3H1	Osteogenesis imperfecta, type VIII, 610915	610339	72	100	100	100
P3H2	Myopia, high, with cataract and vitreoretinal degeneration, 614292	610341	47	100	97	80
PABPN1	Oculopharyngeal muscular dystrophy, 164300	602279	69	98	84	73
PACS1	Schuurs-Hoeijmakers syndrome, 615009	607492	58	100	97	88
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	62	100	98	88
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	45	100	97	82
PAK3	Mental retardation 30/47, 300558	300142	41	100	94	69
PALB2	{Breast cancer, susceptibility to}, 114480 Fanconi anemia, complementation group N, 610832 {Pancreatic cancer, susceptibility to, 3}, 613348	610355	53	100	98	91
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	57	100	100	92
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847	603005	64	100	97	87
PARK7	Parkinson disease 7 early-onset, 606324	602533	55	100	96	78
PAX2	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330	167409	78	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PAX3	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820	606597	77	100	100	100
PAX4	{Diabetes mellitus, ketosis-prone, susceptibility to}, 612227 Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225	167413	53	100	100	96
PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	50	100	98	84
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	167415	76	100	97	93
PAX9	Tooth agenesis, selective, 3, 604625	167416	104	100	100	97
PC	Pyruvate carboxylase deficiency, 266150	608786	90	100	100	100
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070	126090	48	100	94	94
PCCA	Propionicacidemia, 606054	232000	50	100	99	85
PCCB	Propionicacidemia, 606054	232050	53	100	96	86
PCDH15	Deafness 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083	605514	55	100	98	87
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	92	100	99	92
PCM1	No OMIM phenotype	600299	59	100	98	87
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	84	100	99	95
PCSK1	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362	162150	48	100	97	80
PCSK9	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776	607786	70	100	96	91
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	57	100	90	74
PDCD10	Cerebral cavernous malformations 3, 603285	609118	40	100	96	70
PDE11A	Pigmented nodular adrenocortical disease, primary, 2, 610475	604961	54	100	95	78
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799	600129	50	100	96	81
PDE6A	Retinitis pigmentosa 43, 613810	180071	50	100	98	84

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PDE6B	Night blindness, congenital stationary 2, 163500 Retinitis pigmentosa-40, 613801	180072	75	100	100	98
PDE6C	Cone dystrophy 4, 613093	600827	48	100	99	87
PDE6G	Retinitis pigmentosa 57, 613582	180073	91	100	100	100
PDE6H	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024	601190	39	100	100	74
PDE8B	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, 609161	603390	48	100	97	87
PDGFB	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174	190040	63	100	100	100
PDGFRA	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685	173490	55	99	96	87
PDGFRB	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 (4) Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812	173410	78	100	100	99
PDGFRL	Colorectal cancer, somatic, 114500 Hepatocellular cancer, somatic, 114550	604584	92	100	100	97
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	42	99	93	79
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	44	100	98	82
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	72	100	100	100
PDS5B	No OMIM phenotype	605333	38	100	94	70
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	50	100	93	84
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	48	100	95	78
PDX1	{Diabetes mellitus, type II, susceptibility to}, 125853 MODY, type IV, 606392 Pancreatic agenesis 1, 260370	600733	49	100	100	95
PDYN	Spinocerebellar ataxia 23, 610245	131340	80	100	100	100
PDZD7	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	67	100	100	97
PEPD	Prolidase deficiency, 170100	613230	58	100	98	91
PER2	Advanced sleep phase syndrome, familial, 1, 604348	603426	68	100	99	92
PET100	Mitochondrial complex IV deficiency, 220110	614770	75	100	100	81
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	49	100	98	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	64	100	96	89
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	105	100	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	54	100	100	86
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	55	100	100	93
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887	601791	74	100	100	97
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	74	100	95	91
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	46	100	100	93
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	52	100	100	99
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	76	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	44	100	99	84
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	76	100	100	98
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	62	100	99	91
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	43	100	100	87
PFKM	Glycogen storage disease VII, 232800	610681	61	100	100	94
PFN1	Amyotrophic lateral sclerosis 18, 614808	176610	99	100	100	100
PGAM2	Glycogen storage disease X, 261670	612931	93	100	100	100
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	94	100	100	99
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	62	100	100	98
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	48	100	100	91
PGM1	Congenital disorder of glycosylation, type It, 614921	171900	50	100	99	89
PHEX	Hypophosphatemic rickets dominant, 307800	300550	40	100	93	76
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	48	100	95	73
PHF8	Mental retardation syndrome, Siderius type, 300263	300560	49	100	96	74
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	103	100	100	100
PHKA1	Muscle glycogenosis, 300559	311870	39	100	96	67

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PHKA2	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000	300798	42	100	95	81
PHKB	Phosphorylase kinase deficiency of liver and muscle, 261750	172490	54	100	99	91
PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027	172471	83	100	100	100
PHOX2A	Fibrosis of extraocular muscles, congenital, 2, 602078	602753	41	100	98	76
PHOX2B	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013	603851	97	100	100	100
PHRF1	No OMIM phenotype	611780	84	100	100	97
PHYH	Refsum disease, 266500	602026	80	100	92	78
PICALM	Leukemia, acute myeloid, somatic, 601626	603025	42	100	92	64
PIEZO1	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843	611184	91	100	99	98
PIEZO2	Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 ?Marden-Walker syndrome, 248700	613629	49	100	97	84
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	65	100	100	99
PIGL	CHIME syndrome, 280000	605947	62	100	100	81
PIGM	Glycosylphosphatidylinositol deficiency, 610293	610273	58	100	100	99
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	45	100	93	72
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	83	100	100	99
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	71	100	100	94
PIK3CA	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000	171834	65	100	99	92
PIK3CD	Immunodeficiency 14, 615513	602839	83	100	99	98

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PIK3R1	?Agammaglobulinemia 7, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	59	100	99	89
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	59	96	93	89
PIK3R5	Ataxia-oculomotor apraxia 3, 615217	611317	67	100	100	94
PIKFYVE	Corneal fleck dystrophy, 121850	609414	49	100	98	87
PINK1	Parkinson disease 6, early onset, 605909	608309	74	100	97	90
PIP5K1C	Lethal congenital contractural syndrome 3, 611369	606102	77	95	95	95
PITPNM3	Cone-rod dystrophy 5, 600977	608921	66	99	99	97
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550 (4)	602149	125	100	100	97
PITX2	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550	601542	98	100	100	100
PITX3	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623	602669	97	100	100	100
PKD1	Polycystic kidney disease, adult type I, 173900	601313	69	97	94	88
PKD2	Polycystic kidney disease 2, 613095	173910	45	100	96	72
PKHD1	Polycystic kidney and hepatic disease, 263200	606702	58	100	98	89
PKLR	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200	609712	92	100	100	100
PKP1	Ectodermal dysplasia/skin fragility syndrome, 604536	601975	68	100	96	91
PKP2	Arrhythmogenic right ventricular dysplasia 9, 609040	602861	95	100	98	96
PLA2G4A	Phospholipase A2, group IV A, deficiency of	600522	50	100	99	89
PLA2G5	[Fleck retina, familial benign], 228980	601192	56	100	100	94
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	80	100	100	99
PLA2G7	{Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050 Platelet-activating factor acetylhydrolase deficiency, 614278	601690	73	100	99	86
PLAG1	Adenomas, salivary gland pleomorphic, somatic, 181030	603026	63	100	100	100
PLAU	{Alzheimer disease, late-onset, susceptibility to}, 104300 Quebec platelet disorder, 601709	191840	102	100	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	44	100	96	81
PLCB4	Auriculocondylar syndrome 2, 614669	600810	42	100	96	76
PLCD1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600	602142	80	100	100	100

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PLCE1	Nephrotic syndrome, type 3, 610725	608414	56	100	99	93
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468	600220	70	100	99	92
PLEC	Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, type 2Q, 613723	601282	87	100	100	99
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, 4, 611067	611101	67	100	100	96
PLEKHM1	Osteopetrosis 6, 611497	611466	134	100	100	100
PLG	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090	173350	58	100	100	94
PLIN1	Lipodystrophy, familial partial, type 4, 613877	170290	62	100	100	99
PLN	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874	172405	90	100	100	100
PLOD1	Ehlers-Danlos syndrome, type VI, 225400	153454	57	100	100	97
PLOD2	Bruck syndrome 2, 609220	601865	44	100	95	77
PLOD3	Lysyl hydroxylase 3 deficiency, 612394	603066	68	100	100	99
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	74	100	99	87
PLS3	Bone mineral density QTL18, osteoporosis, 300910	300131	38	100	94	76
PML	Leukemia, acute promyelocytic, PML/RARA type	102578	79	100	100	98
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	53	100	100	91
PMP22	Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 ?Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800	601097	74	100	100	97
PMS2	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300	600259	106	100	100	99
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	72	100	100	97
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	54	100	97	89
PNPLA1	Ichthyosis, congenital 10, 615024	612121	68	100	100	96
PNPLA2	Neutral lipid storage disease with myopathy, 610717	609059	70	100	99	96

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PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	78	100	100	98
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	53	100	100	85
PNPT1	Combined oxidative phosphorylation deficiency 13, 614932 Deafness 70, 614934	610316	43	100	94	69
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	55	100	100	98
POF1B	?Premature ovarian failure 2B, 300604	300603	41	100	95	79
POFUT1	Dowling-Degos disease 2, 615327	607491	64	100	100	99
POGLUT1	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, type 2Z, 617232	615618	39	100	98	82
POLD1	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381	174761	67	100	97	93
POLE	{Colorectal cancer, susceptibility to, 12}, 615083 FILS syndrome, 615139	174762	78	100	100	96
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	71	100	100	99
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions 4, 610131	604983	68	100	99	91
POLH	Xeroderma pigmentosum, variant type, 278750	603968	53	100	99	91
POLR1C	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390	610060	76	100	100	91
POLR1D	Treacher Collins syndrome 2, 613717	613715	50	100	100	100
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	60	100	99	89
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	53	100	97	81
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665	176830	84	100	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 Retinitis pigmentosa 76, 617123	606822	63	100	100	95
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830	614828	119	100	100	100
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952	613386	34	100	93	56

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POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	69	100	100	98
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	52	100	99	89
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	124015	84	100	100	100
PORCN	Focal dermal hypoplasia, 305600	300651	74	100	99	93
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	66	100	100	99
POU3F4	Deafness 2, 304400	300039	73	100	100	100
POU4F3	Deafness 15, 602459	602460	123	100	100	100
PPARG	Carotid intimal medial thickness 1, 609338 {Diabetes, type 2}, 125853 Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 [Obesity, resistance to] Obesity, severe, 601665	601487	53	100	100	94
PPIB	Osteogenesis imperfecta, type IX, 259440	123841	52	100	100	92
PPM1D	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450	605100	67	100	100	96
PPM1K	?Maple syrup urine disease, mild variant, 615135	611065	52	100	100	85
PPOX	Porphyria variegata, 176200	600923	73	100	100	98
PPP1CB	No OMIM phenotype	600590	43	100	99	81
PPP1R3A	Insulin resistance, severe, digenic, 125853	600917	56	100	100	99
PPP2R1B	Lung cancer, 211980	603113	53	100	98	86
PPP2R2B	Spinocerebellar ataxia 12, 604326	604325	51	100	98	86
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	51	100	100	87
PQBP1	Renpenning syndrome, 309500	300463	92	100	100	100
PRCC	Renal cell carcinoma, papillary, 605074	179755	62	100	98	91
PRCD	Retinitis pigmentosa 36, 610599	610598	50	100	100	98
PRDM16	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373	605557	92	100	100	98
PRDM5	Brittle cornea syndrome 2, 614170	614161	54	100	99	87
PRELID2	No OMIM phenotype	No id	40	100	93	73
PRF1	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027	170280	87	100	100	100

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PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250	604283	65	100	94	81
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437	608500	61	100	100	95
PRICKLE2	No OMIM phenotype	608501	85	100	100	100
PRIMPOL	Myopia 22, 615420	615421	39	94	86	67
PRKAG2	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200	602743	86	100	100	93
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489	188830	64	100	100	94
PRKCA	Pituitary tumor, invasive	176960	77	100	100	95
PRKCG	Spinocerebellar ataxia 14, 605361	176980	70	100	100	94
PRKCSH	Polycystic liver disease 1, 174050	177060	72	100	100	99
PRKG1	Aortic aneurysm, familial thoracic 8, 615436	176894	45	100	99	88
PRKN	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 {Leprosy, susceptibility to}, 607572 Parkinson disease, juvenile, type 2, 600116	602544	57	100	100	93
PRKRA	Dystonia 16, 612067	603424	67	100	99	95
PRLR	?Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554	176761	52	100	99	91
PRNP	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 {Kuru, susceptibility to}, 245300 Prion disease with protracted course, 606688	176640	109	100	100	100
PROC	Thrombophilia due to protein C deficiency, 176860 Thrombophilia due to protein C deficiency, 612304	612283	86	100	100	98
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	60	100	92	81
PROK2	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628	607002	38	100	100	81
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200	607123	138	100	100	100

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PROM1	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786	604365	57	100	96	74
PROP1	Pituitary hormone deficiency, combined, 2, 262600	601538	69	100	100	97
PROS1	Thrombophilia due to protein S deficiency, 612336 Thrombophilia due to protein S deficiency, 614514	176880	43	98	85	66
PRPF3	Retinitis pigmentosa 18, 601414	607301	58	100	97	90
PRPF31	Retinitis pigmentosa 11, 600138	606419	66	100	100	94
PRPF6	Retinitis pigmentosa 60, 613983	613979	76	100	100	95
PRPF8	Retinitis pigmentosa 13, 600059	607300	71	100	99	94
PRPH2	Choriodal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic, 608133 Retinitis punctata albescens, 136880	179605	94	100	100	100
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	43	100	96	79
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751	614386	75	100	100	100
PRRX1	Agnathia-otocephaly complex, 202650	167420	67	100	100	99
PRSS1	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044	276000	142	100	100	100
PRSS12	Mental retardation 1, 249500	606709	59	100	100	93
PRSS56	Microphthalmia, isolated 6, 613517	613858	58	100	100	99
PRX	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900	605725	122	100	100	99
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	77	100	97	92
PSAT1	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	610936	50	100	100	91

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PSENEN	Acne inversa, familial, 2, 613736	607632	76	100	100	100
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040	177046	82	100	100	93
PSMC3IP	Ovarian dysgenesis 3, 614324	608665	73	100	100	100
PSPH	Phosphoserine phosphatase deficiency, 614023	172480	37	100	90	61
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416	606347	71	100	100	95
PTCH1	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828	601309	70	100	97	92
PTCH2	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255	603673	84	100	100	100
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	44	100	99	84
PTEN	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 {Glioma susceptibility 2}, 613028 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 {Meningioma}, 607174 PTEN hamartoma tumor syndrome {Prostate cancer, somatic}, 176807 Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950	601728	85	92	81	77
PTF1A	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069	607194	78	100	100	97
PTGIS	Hypertension, essential, 145500	601699	68	100	94	93
PTH	Hypoparathyroidism, 146200 Hypoparathyroidism, 146200	168450	65	100	96	86
PTH1R	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400	168468	80	100	100	99
PTHLH	Brachydactyly, type E2, 613382 Humoral hypercalcemia of malignancy	168470	87	100	100	98

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PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	57	100	96	80
PTPN12	Colon cancer, somatic, 114500	600079	44	100	99	87
PTPN14	Choanal atresia and lymphedema, 613611	603155	78	100	99	93
PTPRC	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971	151460	53	100	98	85
PTPRJ	Colon cancer, somatic, 114500	600925	60	97	96	84
PTPRO	Nephrotic syndrome, type 6, 614196	600579	53	100	99	90
PTPRQ	Deafness 84A, 613391	603317	61	100	98	90
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	54	100	100	79
PUF60	Verheij syndrome, 615583	604819	87	100	100	98
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	74	100	99	92
PYCR1	Cutis laxa, type IIB, 612940 Cutis laxa, type IIIB, 614438	179035	66	100	100	95
PYGL	Glycogen storage disease VI, 232700	613741	57	100	98	82
PYGM	McArdle disease, 232600	608455	75	100	100	100
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	75	100	100	96
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	52	100	97	84
RAB18	Warburg micro syndrome 3, 614222	602207	71	100	100	88
RAB23	Carpenter syndrome, 201000	606144	71	100	100	97
RAB27A	Griscelli syndrome, type 2, 607624	603868	37	100	94	71
RAB28	Cone-rod dystrophy 18, 615374	612994	41	100	100	82
RAB33B	Smith-McCort dysplasia 2, 615222	605950	53	100	100	100
RAB39B	Mental retardation 72, 300271 ?Waisman syndrome, 311510	300774	50	100	100	96
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	49	100	99	89
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	49	100	96	79
RAB40AL	No OMIM phenotype	300405	173	100	100	100
RAB7A	Charcot-Marie-Tooth disease, type 2B, 600882	602298	66	100	100	90
RAC2	Neutrophil immunodeficiency syndrome, 608203	602049	64	100	100	100
RAD21	Cornelia de Lange syndrome 4, 614701	606462	53	100	99	83
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	71	100	100	95
RAD51	{Breast cancer, susceptibility to}, 114480 ?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508	179617	36	88	87	67

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RAD51C	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 Fanconi anemia, complementation group O, 613390	602774	49	100	98	83
RAD54B	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027	604289	71	100	98	89
RAD54L	Adenocarcinoma, colonic, somatic {Breast cancer, invasive ductal}, 114480 Lymphoma, non-Hodgkin, somatic, 605027	603615	69	100	100	95
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	54	100	95	87
RAG1	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 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457	179615	75	100	100	100
RAG2	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457	179616	61	100	100	100
RAI1	Smith-Magenis syndrome, 182290	607642	99	100	100	100
RAP1GDS1	Lymphocytic leukemia, acute T-cell	179502	52	100	98	85
RAPSN	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326	601592	73	100	100	96
RARB	Microphthalmia, syndromic 12, 615524	180220	54	100	100	92
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	47	100	97	76
RASA1	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation, 608354 Parkes Weber syndrome, 608355	139150	50	100	95	74
RAX2	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757	610362	63	100	100	100
RB1	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280	614041	104	100	100	99
RB1CC1	Breast cancer, somatic, 114480	606837	55	100	98	87
RBBP8	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744	604124	44	100	98	80

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RBM10	TARP syndrome, 311900	300080	61	100	95	91
RBM20	Cardiomyopathy, dilated, 1DD, 613172	613171	78	100	100	96
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	612074	47	100	99	85
RBM8A	Thrombocytopenia-absent radius syndrome, 274000	605313	82	100	100	100
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	71	100	100	100
RBPJ	Adams-Oliver syndrome 3, 614814	147183	59	100	95	78
RD3	Leber congenital amaurosis 12, 610612	180040	82	100	100	100
RDH12	Leber congenital amaurosis 13, 612712	608830	76	100	100	95
RDH5	Fundus albipunctatus, 136880	601617	81	100	100	100
RDX	Deafness 24, 611022	179410	49	100	94	75
RECQL4	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400	603780	95	100	100	98
REEP1	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, 610250	609139	50	100	100	89
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	52	100	98	85
REN	[Hyperproreninemia] Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430	179820	65	100	100	97
RET	Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 1}, 142623 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300	164761	92	100	100	99
RETREG1	Neuropathy, hereditary sensory and autonomic, type IIB, 613115	613114	47	100	96	77
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	43	100	96	79
RFTN2	No OMIM phenotype	No id	50	100	97	84
RFX5	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920	601863	72	100	100	95
RFX6	Mitchell-Riley syndrome, 615710	612659	62	100	97	80
RFXANK	MHC class II deficiency, complementation group B, 209920	603200	71	100	100	99
RFXAP	Bare lymphocyte syndrome, type II, complementation group D, 209920	601861	129	100	100	100
RGR	Retinitis pigmentosa 44, 613769	600342	65	100	100	96
RGS9	Bradyopsia, 608415	604067	65	100	98	85
RGS9BP	Bradyopsia, 608415	607814	92	100	100	100

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RHAG	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000 Rh-mod syndrome	180297	42	100	97	76
RHBDF2	Tylosis with esophageal cancer, 148500	614404	77	100	100	100
RHCE	[Blood group, Rhesus], 111690 Rh-null disease, amorph type	111700	133	100	100	96
RHO	Night blindness, congenital stationary 1, 610445 Retinitis pigmentosa 4 or recessive, 613731 Retinitis punctata albescens, 136880	180380	78	100	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	63	100	99	91
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075	610222	65	100	99	90
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650	605706	100	100	100	100
RIT1	Noonan syndrome 8, 615355	609591	53	100	100	94
RLBP1	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880	180090	72	100	100	92
RMND1	Combined oxidative phosphorylation deficiency 11, 614922	614917	50	100	96	70
RMRP	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460	157660	No coverage data			
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	73	100	100	99
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	48	100	97	81
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	139	100	100	100
RNASEL	Prostate cancer 1, 601518	180435	59	100	100	97
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	68	100	100	84
RNF123	No OMIM phenotype	614472	74	100	100	99
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	59	100	100	97
RNF139	Renal cell carcinoma, 144700	603046	71	100	100	100
RNF145	No OMIM phenotype	No id	37	99	82	54
RNF168	RIDDLE syndrome, 611943	612688	70	100	98	90
RNF170	Ataxia, sensory, 1, 608984	614649	56	100	94	72
RNF212	Recombination rate QTL 1, 612042	612041	62	100	98	82
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	609948	54	100	92	75
RNF6	Esophageal carcinoma, somatic, 133239	604242	59	100	100	97
ROBO2	Vesicoureteral reflux 2, 610878	602431	54	100	99	91
ROBO3	Gaze palsy, horizontal, with progressive scoliosis, 607313	608630	76	100	100	99
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	76	100	97	94

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ROM1	Retinitis pigmentosa 7, digenic, 608133	180721	74	100	100	100
ROR2	Brachydactyly, type B1, 113000 Robinow syndrome, 268310	602337	106	100	100	98
RP1	Retinitis pigmentosa 1, 180100	603937	60	100	100	96
RP1L1	Occult macular dystrophy, 613587	608581	98	100	100	100
RP2	Retinitis pigmentosa 2, 312600	300757	49	100	100	93
RPE65	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794	180069	52	100	100	94
RPGR	Cone-rod dystrophy, 1, 304020 Macular degeneration atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, and sinorespiratory infections, with or without deafness, 300455	312610	37	82	70	59
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	59	100	98	86
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	47	97	94	77
RPIA	?Ribose 5-phosphate isomerase deficiency, 608611	180430	52	100	98	89
RPL11	Diamond-Blackfan anemia 7, 612562	604175	44	100	100	94
RPL35A	Diamond-Blackfan anemia 5, 612528	180468	73	100	100	100
RPL5	Diamond-Blackfan anemia 6, 612561	603634	50	100	96	84
RPS10	Diamond-Blackfan anemia 9, 613308	603632	46	100	98	86
RPS14	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550	130620	77	100	100	100
RPS17	Diamond-Blackfan anemia 4, 612527	180472	74	100	100	90
RPS19	Diamond-Blackfan anemia 1, 105650	603474	72	100	100	95
RPS24	Diamond-blackfan anemia 3, 610629	602412	76	100	100	99
RPS26	Diamond-Blackfan anemia 10, 613309	603701	70	100	100	98
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation 19, 300844	300075	38	100	85	57
RPS7	Diamond-Blackfan anemia 8, 612563	603658	49	100	100	91
RPSA	Asplenia, isolated congenital, 271400	150370	72	100	100	100
RRAS2	Ovarian carcinoma	600098	48	100	96	77
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions 5, 613077	604712	60	100	100	95
RS1	Retinoschisis, 312700	300839	38	100	92	69
RSPH1	Ciliary dyskinesia, primary, 24, 615481	609314	45	100	96	79
RSPH4A	Ciliary dyskinesia, primary, 11, 612649	612647	73	100	100	96

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RSPH9	Ciliary dyskinesia, primary, 12, 612650	612648	85	100	100	99
RSPO1	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644	609595	50	100	100	94
RSPO4	Anonychia congenita, 206800	610573	63	100	100	100
RTEL1	Dyskeratosis congenita 4, 615190 Dyskeratosis congenita 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	80	100	100	98
RTN2	Spastic paraplegia 12, 604805	603183	100	100	100	100
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833	610436	49	100	95	79
RUNX1	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399	151385	71	100	100	97
RUNX2	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510	600211	66	100	100	95
RXFP2	No OMIM phenotype	606655	41	100	96	75
RYR1	Central core disease, 117000 King-Denborough syndrome, 145600 {Malignant hyperthermia susceptibility 1}, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000	180901	90	99	99	98
RYR2	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772	180902	85	100	98	90
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550	604490	58	100	100	98
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	59	100	99	84
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480	602218	99	100	100	100
SALL4	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750	607343	115	100	98	95
SAMD9	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455	610456	64	100	100	100
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	49	100	93	69
SAR1B	Chylomicron retention disease, 246700	607690	53	100	100	82
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845	612804	65	100	100	98
SART3	No OMIM phenotype	611684	61	100	98	86
SAT1	No OMIM phenotype	313020	57	100	95	84

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SATB2	Glass syndrome, 612313	608148	69	100	98	87
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	49	100	100	92
SBF2	Charcot-Marie-Tooth disease, type 4B2, 604563	607697	52	100	98	84
SC5D	Lathosterolosis, 607330	602286	68	100	100	99
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	602257	60	100	98	88
SCARF2	Van den Ende-Gupta syndrome, 600920	613619	75	99	97	93
SCN10A	Episodic pain syndrome, familial, 2, 615551	604427	77	100	100	96
SCN11A	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548	604385	62	100	99	92
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6, 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634	182389	67	100	100	94
SCN1B	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350	600235	105	100	93	93
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	73	100	99	95
SCN2B	Atrial fibrillation, familial, 14, 615378	601327	68	100	100	100
SCN3B	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120	608214	59	100	100	100
SCN4A	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300	603967	95	100	100	99
SCN4B	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819	608256	68	100	100	98

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SCN5A	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 {Sudden infant death syndrome, susceptibility to}, 272120 Ventricular fibrillation, familial, 1, 603829	600163	121	100	100	100
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	72	100	99	91
SCN9A	{Dravet syndrome, modifier of}, 607208 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder,, 167400 Small fiber neuropathy, 133020	603415	63	100	99	94
SCNN1A	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350	600228	80	100	100	95
SCNN1B	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350	600760	70	100	100	94
SCNN1G	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350	600761	72	100	98	92
SCO1	Mitochondrial complex IV deficiency, 220110	603644	61	100	100	92
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	81	100	100	100
SCP2	?Leukoencephalopathy with dystonia and motor neuropathy, 613724	184755	40	100	98	78
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	66	100	99	87
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	93	100	93	83

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SDHAF1	Mitochondrial complex II deficiency, 252011	612848	51	100	100	100
SDHAF2	Paragangliomas 2, 601650	613019	91	100	100	93
SDHB	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300	185470	80	100	100	100
SDHC	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373	602413	113	100	100	100
SDHD	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300	602690	111	100	100	100
SEC23A	Craniolenticulosutural dysplasia, 607812	610511	44	100	95	77
SEC23B	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100	610512	52	100	96	84
SEC63	Polycystic liver disease 2, 617004	608648	42	100	96	75
SECISBP2	Thyroid hormone metabolism, abnormal, 609698	607693	59	100	99	87
SELENON	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310	606210	74	90	90	88
SEMA3E	?CHARGE syndrome, 214800	608166	44	100	98	78
SEMA4A	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282	607292	72	100	100	99
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	49	100	96	77
SEPT12	Spermatogenic failure 10, 614822	611562	66	100	99	92
SEPT9	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related Ovarian carcinoma	604061	66	100	100	97
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	44	100	92	66
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	65	100	100	93

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SERPINA3	Alpha-1-antichymotrypsin deficiency Cerebrovascular disease, occlusive	107280	71	100	100	98
SERPINA6	Corticosteroid-binding globulin deficiency, 611489	122500	76	100	100	100
SERPINA7	[Thyroxine-binding globulin QTL], 300932	314200	45	100	96	79
SERPINB6	?Deafness 91, 613453	173321	60	100	98	82
SERPINB7	Palmoplantar keratoderma, Nagashima type, 615598	603357	51	100	100	89
SERPINC1	Thrombophilia due to antithrombin III deficiency, 613118	107300	65	100	100	93
SERPIND1	Thrombophilia due to heparin cofactor II deficiency, 612356	142360	53	100	100	95
SERPINE1	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}	173360	82	100	100	98
SERPINF1	Osteogenesis imperfecta, type VI, 613982	172860	68	100	99	86
SERPINF2	Alpha-2-plasmin inhibitor deficiency, 262850	613168	80	100	100	100
SERPING1	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790	606860	74	100	100	96
SERPINH1	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504	600943	95	100	100	100
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218	602445	55	100	100	91
SETBP1	Mental retardation 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	70	98	96	94
SETD1A	No OMIM phenotype	611052	71	99	96	93
SETD5	Mental retardation 23, 615761	615743	57	100	99	90
SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia 1, 606002	608465	55	100	99	92
SF3B1	Myelodysplastic syndrome, somatic, 614286	605590	52	100	99	86
SF3B4	Acrofacial dysostosis 1, Nager type, 154400	605593	72	100	100	100
SFTPA2	Pulmonary fibrosis, idiopathic, 178500	178642	127	100	100	100
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1, 265120	178640	64	100	100	100
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2, 610913	178620	62	100	100	100
SFXN4	Combined oxidative phosphorylation deficiency 18, 615578	615564	50	100	98	86
SGCA	Muscular dystrophy, limb-girdle, type 2D, 608099	600119	84	100	100	100
SGCB	Muscular dystrophy, limb-girdle, type 2E, 604286	600900	39	100	93	75
SGCD	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287	601411	41	100	99	84
SGCE	Dystonia-11, myoclonic, 159900	604149	56	100	98	86
SGCG	Muscular dystrophy, limb-girdle, type 2C, 253700	608896	53	100	96	77
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	90	100	96	94

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SH2B3	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950	605093	76	100	100	100
SH2D1A	Lymphoproliferative syndrome, 1, 308240	300490	44	100	99	79
SH3BP2	Cherubism, 118400	602104	62	91	91	91
SH3PXD2B	Frank-ter Haar syndrome, 249420	613293	93	100	100	97
SH3TC2	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353	608206	63	100	100	95
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	62	93	78	72
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	68	100	98	95
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	48	100	98	85
SHOX	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582	312865	38	100	96	71
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434	300579	61	100	98	96
SI	Sucrase-isomaltase deficiency, congenital, 222900	609845	45	100	97	80
SIGMAR1	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, 2, 605726	601978	74	100	100	100
SIL1	Marinesco-Sjogren syndrome, 248800	608005	54	100	100	93
SIM1	Obesity, severe, 601665	603128	66	100	100	97
SIX1	Branchiootic syndrome 3, 608389 Deafness 23, 605192	601205	84	100	100	97
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	99	100	100	100
SIX5	Branchiootorenal syndrome 2, 610896	600963	66	100	100	93
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, 212550	606326	131	100	100	100
SKI	Shprintzen-Goldberg syndrome, 182212	164780	76	100	100	99
SKIV2L	Trichohepatoenteric syndrome 2, 614602	600478	87	100	100	100
SLC10A2	Bile acid malabsorption, primary, 613291	601295	61	100	98	85
SLC11A2	Anemia, hypochromic microcytic, with iron overload 1, 206100	600523	38	100	90	64
SLC12A1	Bartter syndrome, type 1, 601678	600839	53	100	99	86
SLC12A3	Gitelman syndrome, 263800	600968	69	100	100	98
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	46	100	97	83

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SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095	600682	61	100	100	96
SLC16A12	Cataract 47, juvenile, with microcornea, 612018	611910	63	100	100	96
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	60	100	100	86
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	60	100	99	83
SLC17A8	Deafness 25, 605583	607557	48	100	100	93
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270	603941	50	100	99	89
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	54	100	100	94
SLC1A3	Episodic ataxia, type 6, 612656	600111	66	100	100	97
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600	158378	65	100	98	86
SLC22A12	Hypouricemia, renal, 220150	607096	68	100	100	96
SLC22A18	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210	602631	62	100	97	89
SLC22A5	Carnitine deficiency, systemic primary, 212140	603377	72	100	100	97
SLC24A1	Night blindness, congenital stationary (complete), 1D, 613830	603617	68	100	100	97
SLC24A5	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750	609802	64	100	100	98
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182	190315	66	100	100	100
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	61	100	100	91
SLC25A13	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814	603859	72	100	94	81
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	85	100	100	94
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	70	100	100	97
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138	613698	42	100	98	80
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	81	100	100	100
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773	600370	73	100	100	97
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950	610819	83	100	100	100
SLC25A4	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions 2, 609283	103220	74	100	100	100

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SLC26A2	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900	606718	54	100	100	99
SLC26A3	Diarrhea 1, secretory chloride, congenital, 214700	126650	52	100	99	88
SLC26A4	Deafness 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600	605646	47	100	97	78
SLC26A5	?Deafness 61, 613865	604943	43	100	98	84
SLC26A8	Spermatogenic failure 3, 606766	608480	46	100	97	80
SLC27A4	Ichthyosis prematurity syndrome, 608649	604194	78	100	100	100
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, 602782	612373	85	100	100	99
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	94	100	100	100
SLC2A10	Arterial tortuosity syndrome, 208050	606145	94	100	100	96
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi-Bickel syndrome, 227810	138160	47	100	99	86
SLC2A9	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076	606142	69	100	100	95
SLC30A10	Hypermanganesemia with dystonia 1, 613280	611146	81	100	100	98
SLC30A2	Zinc deficiency, transient neonatal, 608118	609617	66	100	100	98
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	55	100	97	78
SLC34A1	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286	182309	78	100	100	100
SLC34A2	Pulmonary alveolar microlithiasis, 265100	604217	78	100	100	94
SLC34A3	Hypophosphatemic rickets with hypercalciuria, 241530	609826	75	96	94	91
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	49	100	99	82
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	55	100	100	98
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	89	100	100	99
SLC35D1	Schneckenbecken dysplasia, 269250	610804	42	100	90	64
SLC36A2	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608331	69	100	100	99

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SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	69	100	100	97
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	615585	52	100	99	89
SLC39A13	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350	608735	89	100	100	100
SLC39A4	Acrodermatitis enteropathica, 201100	607059	71	100	100	100
SLC3A1	Cystinuria, 220100	104614	63	100	99	91
SLC40A1	Hemochromatosis, type 4, 606069	604653	47	100	100	89
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	60	100	100	89
SLC46A1	Folate malabsorption, hereditary, 229050	611672	75	100	100	99
SLC4A1	[Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 Cryohydrocytosis, 185020 [Malaria, resistance to], 611162 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653	109270	81	100	100	100
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, 217700	610206	86	100	100	99
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	603345	43	100	99	83
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500	613350	69	100	100	100
SLC5A1	Glucose/galactose malabsorption, 606824	182380	70	100	96	86
SLC5A2	Renal glucosuria, 233100	182381	78	100	100	99
SLC5A5	Thyroid dyshormonogenesis 1, 274400	601843	63	100	100	97
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580	608761	59	100	100	92
SLC6A19	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608893	73	100	100	98
SLC6A2	Orthostatic intolerance, 604715	163970	64	100	100	96

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SLC6A20	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	605616	80	100	100	99
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	65	100	100	99
SLC6A5	Hyperekplexia 3, 614618	604159	58	100	99	87
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	79	100	97	92
SLC7A14	Retinitis pigmentosa 68, 615725	615720	76	100	100	97
SLC7A7	Lysinuric protein intolerance, 222700	603593	54	100	97	88
SLC7A9	Cystinuria, 220100	604144	62	100	100	96
SLC9A3R1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287	604990	74	100	100	99
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	54	100	94	82
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic, 237450	604843	51	100	97	79
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic, 237450	605495	54	100	96	85
SLCO2A1	Hypertrophic osteoarthropathy, primary 2, 614441	601460	67	100	100	98
SLITRK1	Tourette syndrome, 137580 ?Trichotillomania, 613229	609678	68	100	100	99
SLITRK6	Deafness and myopia, 221200	609681	65	100	100	98
SLURP1	Meleda disease, 248300	606119	79	100	100	100
SLX4	Fanconi anemia, complementation group P, 613951	613278	87	100	100	98
SMAD3	Loeys-Dietz syndrome 3, 613795	603109	99	100	100	100
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	59	100	100	96
SMAD6	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439	602931	90	100	100	99
SMAD9	Pulmonary hypertension, primary, 2, 615342	603295	67	100	100	98
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	61	98	96	85
SMARCA4	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325	603254	76	100	100	98
SMARCAD1	Adermatoglyphia, 136000 Basan syndrome, 129200	612761	48	100	95	77
SMARCAL1	Schimke immunoosseous dysplasia, 242900	606622	61	100	99	93
SMARCB1	Coffin-Siris syndrome 3, 614608 {Rhabdoid predisposition syndrome 1}, 609322 Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091	601607	71	100	100	95
SMC1A	Cornelia de Lange syndrome 2, 300590	300040	59	100	98	95

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SMC3	Cornelia de Lange syndrome 3, 610759	606062	56	100	97	83
SMCHD1	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901	614982	52	100	96	78
SMN1	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150	600354	112	100	100	100
SMO	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707	601500	76	100	100	99
SMOC1	Microphthalmia with limb anomalies, 206920	608488	68	100	98	91
SMOC2	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400	607223	49	100	95	80
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	87	100	99	97
SMPX	Deafness 4, 300066	300226	41	100	96	67
SMS	Mental retardation, Snyder-Robinson type, 309583	300105	41	95	80	70
SNAI2	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890	602150	52	100	100	99
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	91	100	100	96
SNCA	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543	163890	34	100	99	69
SNCB	Dementia, Lewy body, 127750	602569	73	100	100	100
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501	608241	64	100	100	97
SNRNP200	Retinitis pigmentosa 33, 610359	601664	72	100	99	94
SNRPE	Hypotrichosis 11, 615059	128260	38	100	93	58
SNRPN	Prader-Willi syndrome, 176270	182279	64	100	100	100
SNTA1	Long QT syndrome 12, 612955	601017	80	98	87	80
SNX10	Osteopetrosis 8, 615085	614780	56	100	100	92
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671	613667	64	98	95	84
SOD1	Amyotrophic lateral sclerosis 1, 105400	147450	63	100	100	95
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	57	100	98	84
SOST	Craniodiaphyseal dysplasia, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100	605740	115	100	100	100
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	68	100	100	100

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SOX17	Vesicoureteral reflux 3, 613674	610928	85	100	100	100
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940	601618	44	93	83	77
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	100	100	100	100
SOX3	Mental retardation, with isolated growth hormone deficiency, 300123 Panhypopituitarism, 312000	313430	56	100	96	89
SOX9	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290	608160	66	100	100	99
SP110	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948	604457	44	100	99	85
SP7	?Osteogenesis imperfecta, type XII, 613849	606633	92	100	100	100
SPAG1	Ciliary dyskinesia, primary, 28, 615505	603395	45	100	95	73
SPART	Troyer syndrome, 275900	607111	55	100	100	91
SPAST	Spastic paraplegia 4, 182601	604277	46	100	98	80
SPATA16	?Spermatogenic failure 6, 102530	609856	46	100	99	84
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	609868	51	100	98	89
SPECC1L	?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410	614140	45	100	96	80
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	56	100	98	92
SPG21	Mast syndrome, 248900	608181	43	100	91	66
SPG7	Spastic paraplegia 7, 607259	602783	80	100	99	91
SPINK1	{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189	167790	70	100	100	83
SPINK5	Atopy, 147050 Netherton syndrome, 256500	605010	46	100	96	82
SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420	605124	59	100	100	99
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	66	100	100	96
SPRED1	Legius syndrome, 611431	609291	44	100	95	84
SPRY4	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266	607984	122	100	97	96
SPTA1	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970	182860	48	100	98	82

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SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	64	100	99	94
SPTB	Anemia, neonatal hemolytic, fatal and near-fatal Elliptocytosis-3 Spherocytosis, type 2, 616649	182870	76	100	100	97
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia 14, 615386	604985	73	100	100	100
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA, 162400	605712	45	100	97	83
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640	605713	51	100	98	79
SQSTM1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250	601530	76	100	98	90
SRC	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937	190090	65	100	100	92
SRCAP	Floating-Harbor syndrome, 136140	611421	84	100	100	99
SRD5A2	Pseudovaginal perineoscrotal hypospadias, 264600	607306	44	100	94	76
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	63	100	99	92
SRP72	Bone marrow failure syndrome 1, 614675	602122	48	100	98	83
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	47	100	97	85
SRY	46XX sex reversal 1, 400045 46XY sex reversal 1, 400044	480000	20	50	41	35
SSTR5	Somatostatin analog, resistance to	182455	136	100	100	100
ST14	Ichthyosis, congenital 11, 602400	606797	80	100	100	98
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation 12, 611090	606494	52	100	98	87
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	40	94	84	61
STAC3	Native American myopathy, 255995	615521	54	100	98	89
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	51	100	100	90
STAR	Lipoid adrenal hyperplasia, 201710	600617	75	100	100	100
STAT1	Immunodeficiency 31A, mycobacteriosis, 614892 Immunodeficiency 31B, mycobacterial and viral infections, 613796 Immunodeficiency 31C, 614162	600555	43	100	96	80
STAT3	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060	102582	60	100	100	91
STAT5B	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578	604260	71	100	99	89
STIL	Microcephaly 7, primary, 612703	181590	53	100	99	90

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STIM1	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070	605921	64	100	100	94
STK11	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300	602216	83	100	100	100
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868	604965	58	100	100	93
STOX1	Preeclampsia/eclampsia 4, 609404	609397	47	89	89	87
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	69	100	100	99
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	67	100	100	92
STRC	Deafness 16, 603720	606440	120	100	100	100
STS	Ichthyosis, 308100	300747	71	97	97	92
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	53	100	99	86
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, 613101	601717	68	100	100	93
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	46	100	94	80
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	61	100	99	93
SUFU	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174	607035	60	100	100	97
SUMF1	Multiple sulfatase deficiency, 272200	607939	60	100	98	81
SUMO1	Orofacial cleft 10, 613705	601912	57	100	91	81
SUOX	Sulfite oxidase deficiency, 272300	606887	106	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	59	93	87	87
SYCP3	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960	604759	38	100	95	68
SYN1	Epilepsy, with variable learning disabilities and behavior disorders, 300491 dominant	313440	58	100	100	97
SYNE1	Emery-Dreifuss muscular dystrophy 4, 612998 Spinocerebellar ataxia 8, 610743	608441	55	100	99	89
SYNE2	Emery-Dreifuss muscular dystrophy 5, 612999	608442	57	100	98	88
SYNE4	Deafness 76, 615540	615535	74	100	100	100
SYNGAP1	Mental retardation 5, 612621	603384	93	98	98	97
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530	604297	48	100	96	83
SYP	Mental retardation 96, 300802	313475	50	100	98	87

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SYT14	Spinocerebellar ataxia 11, 614229	610949	47	100	93	80
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	77	100	100	98
T	{Neural tube defects, susceptibility to}, 182940 Sacral agenesis with vertebral anomalies, 615709	601397	95	100	100	95
TAB2	Congenital heart defects, nonsyndromic, 2, 614980	605101	58	100	100	95
TAC3	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839	162330	62	100	100	84
TACR3	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840	162332	87	100	100	100
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870	137290	162	100	100	100
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	48	100	95	83
TAF2	Mental retardation 40, 615599	604912	47	100	95	76
TAL1	Leukemia, T-cell acute lymphocytic, somatic, 613065	187040	41	97	87	75
TAL2	Leukemia, T-cell acute lymphocytic, somatic, 613065	186855	62	100	100	100
TALDO1	Transaldolase deficiency, 606003	602063	86	100	100	97
TAP1	Bare lymphocyte syndrome, type I, 604571	170260	83	100	100	99
TAP2	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis	170261	67	100	100	97
TAPBP	Bare lymphocyte syndrome, type I, 604571	601962	70	100	100	100
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069	605078	107	100	100	88
TAT	Tyrosinemia, type II, 276600	613018	48	100	100	91
TAZ	Barth syndrome, 302060	300394	59	100	91	90
TBC1D20	Warburg micro syndrome 4, 615663	611663	53	98	93	93
TBC1D24	DOOR syndrome, 220500 Deafness 86, 614617 Deafness 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	87	100	100	98
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	44	98	90	70
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	46	100	99	81
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430	602054	44	91	78	67
TBX15	Cousin syndrome, 260660	604127	59	100	100	92

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TBX19	Adrenocorticotropic hormone deficiency, 201400	604614	78	100	100	95
TBX20	Atrial septal defect 4, 611363	606061	62	100	97	90
TBX21	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550	604895	96	100	100	100
TBX22	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400	300307	50	100	97	82
TBX3	Ulnar-mammary syndrome, 181450	601621	82	100	100	98
TBX4	Ischiocoxopodopatellar syndrome, 147891	601719	78	100	100	99
TBX5	Holt-Oram syndrome, 142900	601620	72	100	100	94
TBXAS1	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158	274180	50	100	93	81
TCAP	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954	604488	72	100	100	100
TCF12	Craniosynostosis 3, 615314	600480	50	100	99	87
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954	602272	50	100	97	87
TCIRG1	Osteopetrosis 1, 259700	604592	79	100	100	92
TCN2	Transcobalamin II deficiency, 275350	613441	76	100	100	99
TCOF1	Treacher Collins syndrome 1, 154500	606847	78	100	100	98
TCTN1	Joubert syndrome 13, 614173	609863	61	100	99	88
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	613846	60	100	98	90
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	50	100	99	90
TDGF1	Forebrain defects	187395	86	100	100	96
TDP1	Spinocerebellar ataxia with axonal neuropathy, 607250	607198	37	100	91	67
TDRD7	Cataract 36, 613887	611258	50	100	99	89
TEAD1	Sveinsson chorioretinal atrophy, 108985	189967	57	100	100	90
TECPR2	Spastic paraplegia 49, 615031	615000	66	100	99	94
TECR	Mental retardation 14, 614020	610057	81	100	100	94
TECTA	Deafness 8/12, 601543 Deafness 21, 603629	602574	82	100	99	94
TEK	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195	600221	54	100	99	88
TENM3	Microphthalmia, isolated, with coloboma 9, 615145	610083	60	100	99	96
TERC	{Aplastic anemia}, 614743 Dyskeratosis congenita 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743	602322	No coverage data			

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TET2	Myelodysplastic syndrome, somatic, 614286	612839	53	100	100	97
TEX28	No OMIM phenotype	300092	No coverage data			
TF	Atransferrinemia, 209300	190000	50	100	99	89
TFAP2A	Branchiooculofacial syndrome, 113620	107580	80	100	100	100
TFAP2B	Char syndrome, 169100 Patent ductus arteriosus 2, 617035	601601	68	100	100	94
TFE3	Renal cell carcinoma, papillary, 1, 300854	314310	52	100	99	89
TFG	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, 615658	602498	65	100	98	88
TFR2	Hemochromatosis, type 3, 604250	604720	69	100	100	97
TG	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 Thyroid dyshormonogenesis 3, 274700	188450	65	100	100	94
TGFB1	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700	190180	72	100	96	87
TGFB2	Loeys-Dietz syndrome 4, 614816	190220	54	100	100	94
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582	190230	69	100	100	100
TGFBI	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471	601692	52	100	98	85
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	96	95	93	92
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	124	100	100	100
TGIF1	Holoprosencephaly 4, 142946	602630	94	100	100	100
TGM1	Ichthyosis, congenital 1, 242300	190195	79	100	100	99
TGM5	Peeling skin syndrome 2, 609796	603805	78	100	99	93
TGM6	Spinocerebellar ataxia 35, 613908	613900	73	100	100	98
TH	Segawa syndrome, recessive, 605407	191290	66	100	100	97
THAP1	Dystonia 6, torsion, 602629	609520	64	100	100	98
THBD	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 Thrombophilia due to thrombomodulin defect, 614486	188040	131	100	100	100
THOC6	Beaulieu-Boycott-Innes syndrome, 613680	615403	133	100	100	100

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THPO	Thrombocythemia 1, 187950	600044	84	100	100	100
THRA	Hypothyroidism, congenital, nongoitrous, 6, 614450	190120	78	100	100	99
THRB	Thyroid hormone resistance, 188570 Thyroid hormone resistance, 274300 Thyroid hormone resistance, selective pituitary, 145650	190160	56	100	99	93
TIA1	Welander distal myopathy, 604454	603518	55	100	94	78
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	102	100	100	100
TIMP3	Sorsby fundus dystrophy, 136900	188826	65	100	100	100
TINF2	Dyskeratosis congenita 3, 613990 Revesz syndrome, 268130	604319	94	100	100	100
TJP2	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748	607709	57	100	100	92
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 617069	188250	46	100	99	86
TLL1	Atrial septal defect 6, 613087	606742	46	100	97	82
TLR4	No OMIM phenotype	603030	49	100	100	97
TMC1	Deafness 36, 606705 Deafness 7, 600974	606706	43	100	94	77
TMC6	Epidermodysplasia verruciformis, 226400	605828	59	100	100	98
TMC8	Epidermodysplasia verruciformis, 226400	605829	68	100	100	98
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980	614123	76	100	99	86
TMEM126A	Optic atrophy 7, 612989	612988	57	100	100	79
TMEM138	Joubert syndrome 16, 614465	614459	38	100	100	87
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	73	100	100	97
TMEM181	No OMIM phenotype	613209	54	100	99	87
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	61	100	99	76
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	71	100	100	90
TMEM237	Joubert syndrome 14, 614424	614423	38	100	90	69
TMEM38B	Osteogenesis imperfecta, type XIV, 615066	611236	46	100	99	83
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	58	100	98	82
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	78	100	100	94
TMIE	Deafness 6, 600971	607237	57	100	99	99

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TMLHE	{Autism, susceptibility to 6}, 300872	300777	32	100	97	59
TMPRSS15	Enterokinase deficiency, 226200	606635	48	100	97	82
TMPRSS3	Deafness 8/10, 601072	605511	50	100	99	89
TMPRSS6	Iron-refractory iron deficiency anemia, 206200	609862	65	100	100	96
TNC	Deafness 56, 615629	187380	72	100	99	93
TNFRSF10B	Squamous cell carcinoma, head and neck, 275355	603612	62	100	100	96
TNFRSF11A	Osteolysis, familial expansile, 174810 Osteopetrosis 7, 612301 {Paget disease of bone 2, early-onset}, 602080	603499	66	95	95	91
TNFRSF11B	Paget disease of bone 5, juvenile-onset, 239000	602643	61	100	100	96
TNFRSF13B	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529	604907	55	100	100	96
TNFRSF13C	Immunodeficiency, common variable, 4, 613494	606269	53	100	84	65
TNFRSF1A	{Multiple sclerosis, susceptibility to, 5}, 614810 Periodic fever, familial, 142680	191190	66	100	99	93
TNFSF11	Osteopetrosis 2, 259710	602642	42	100	99	73
TNNC1	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243	191040	74	100	100	100
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B, 601680	191043	94	100	100	100
TNNI3	Cardiomyopathy, dilated, 1FF, 613286 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690	191044	93	100	100	100
TNNT1	Nemaline myopathy 5, Amish type, 605355	191041	51	100	97	89
TNNT2	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494	191045	84	100	100	99
TNNT3	Arthyrgryposis, distal, type 2B, 601680	600692	69	100	100	91
TNXB	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963	600985	113	100	100	100
TOP1	DNA topoisomerase I, camptothecin-resistant	126420	56	100	97	81
TOP2A	DNA topoisomerase II, resistance to inhibition of, by amsacrine	126430	56	100	98	91
TOPORS	Retinitis pigmentosa 31, 609923	609507	57	100	100	100

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TP53	Adrenal cortical carcinoma, 202300 {Basal cell carcinoma 7}, 614740 Breast cancer, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 {Glioma susceptibility 1}, 137800 Hepatocellular carcinoma, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, 607107 Osteosarcoma, 259500 Pancreatic cancer, 260350	191170	99	100	100	100
TP63	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 129400 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289	603273	55	100	99	90
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	106	100	100	98
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458	606370	44	100	95	77
TPM1	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878	191010	86	100	100	100
TPM2	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, 609285	190990	67	100	100	99
TPM3	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1 or recessive, 609284	191030	46	100	100	87
TPMT	{Thiopurines, poor metabolism of, 1}, 610460	187680	58	100	87	68
TPO	Thyroid dyshormonogenesis 2A, 274500	606765	70	100	100	98
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	70	100	100	99
TPRN	Deafness 79, 613307	613354	65	90	78	73
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	44	100	95	77
TRAPPC2	Spondyloepiphyseal dysplasia tarda, 313400	300202	47	100	89	67
TRAPPC9	Mental retardation 13, 613192	611966	68	100	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441	603283	42	100	93	70
TREM2	Nasu-Hakola disease, 221770	605086	68	100	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	116	100	100	100
TRHR	Thyrotropin-releasing hormone resistance, generalized	188545	71	100	100	100
TRIM24	No OMIM phenotype	603406	52	100	99	86
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	86	100	100	100
TRIM33	No OMIM phenotype	605769	49	100	96	81
TRIM37	Mulibrey nanism, 253250	605073	44	100	97	80
TRIOBP	Deafness 28, 609823	609761	98	100	100	97
TRIP11	Achondrogenesis, type IA, 200600	604505	57	100	98	89
TRMU	{Deafness, modifier of}, 580000 Liver failure, transient infantile, 613070	610230	67	100	99	89
TRPA1	Episodic pain syndrome, familial, 615040	604775	46	100	96	77
TRPC6	Glomerulosclerosis, focal segmental, 2, 603965	603652	62	100	100	89
TRPM1	Night blindness, congenital stationary (complete), 1C, 613216	603576	60	100	100	95
TRPM4	Progressive familial heart block, type IB, 604559	606936	83	100	100	100
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	55	100	97	89
TRPS1	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351	604386	64	100	100	99
TRPV3	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400	607066	67	100	100	92
TRPV4	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level QTL 1], 613508 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252	605427	73	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	105	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	104	100	100	100
TSEN2	Pontocerebellar hypoplasia type 2B, 612389	608753	45	87	82	67
TSEN34	?Pontocerebellar hypoplasia type 2C, 612390	608754	59	100	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	100	96	95
TSFM	Combined oxidative phosphorylation deficiency 3, 610505	604723	60	100	100	100
TSG101	Breast cancer, somatic, 114480	601387	51	100	98	83
TSHB	Hypothryoidism, congenital, nongoitrous 4, 275100	188540	70	100	100	100
TSHR	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic Thyroid carcinoma with thyrotoxicosis	603372	67	100	99	92
TSHZ1	Aural atresia, congenital, 607842	614427	83	98	98	98
TSPAN12	Exudative vitreoretinopathy 5, 613310	613138	48	100	98	79
TSPAN7	Mental retardation 58, 300210	300096	45	100	95	77
TSPEAR	Deafness 98, 614861	612920	72	100	100	99
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome, 608800	604714	87	100	100	100
TTBK2	Spinocerebellar ataxia 11, 604432	611695	59	100	100	95
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	39	100	89	62
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	612014	56	100	97	84
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	43	100	97	80
TTC7A	Gastrointestinal defects and immunodeficiency syndrome, 243150	609332	68	100	99	94
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	46	100	99	85
TTI2	Mental retardation 39, 615541	614426	46	100	98	81

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TTN	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, early-onset, with fatal cardiomyopathy, 611705 Myopathy, proximal, with early respiratory muscle involvement, 603689 Tibial muscular dystrophy, tardive, 600334	188840	57	100	100	96
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	45	100	96	79
TTR	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680	176300	66	100	100	100
TUBA1A	Lissencephaly 3, 611603	602529	89	100	100	100
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180	605742	79	100	100	100
TUBB1	Macrothrombocytopenia, TUBB1-related, 613112	612901	100	100	100	97
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	170	100	97	95
TUBB2B	Polymicrogyria, symmetric or asymmetric, 610031	612850	170	100	100	100
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	168	100	95	95
TUBB4A	Dystonia 4, torsion, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	148	100	100	97
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	99	100	100	100
TUBGCP6	Microcephaly and chorioretinopathy, 1, 251270	610053	109	100	100	99
TUFM	Combined oxidative phosphorylation deficiency 4, 610678	602389	100	100	100	100
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	69	100	100	98
TUSC3	Mental retardation 7, 611093	601385	55	100	99	88
TWIST1	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400	601622	72	100	90	77
TWIST2	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260	607556	96	100	100	100
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	109	100	100	99
TYK2	Immunodeficiency 35, 611521	176941	83	100	100	100
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	131222	67	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TYR	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 Waardenburg syndrome/albinism, digenic, 103470	606933	67	100	99	93
TYROBP	Nasu-Hakola disease, 221770	604142	71	100	100	100
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	52	100	100	93
UBA1	Spinal muscular atrophy 2, infantile, 301830	314370	70	100	100	98
UBE2A	Mental retardation syndromic, Nascimento-type, 300860	312180	56	100	88	75
UBE3A	Angelman syndrome, 105830	601623	53	100	100	93
UBE3B	Kaufman oculocerebrofacial syndrome, 244450	608047	72	100	100	93
UBIAD1	Corneal dystrophy, Schnyder type, 121800	611632	93	100	100	100
UBQLN2	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857	300264	70	100	100	100
UBR1	Johanson-Blizzard syndrome, 243800	605981	43	100	97	77
UGT1A1	[Bilirubin, serum level of, QTL1], 601816 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 [Gilbert syndrome], 143500 Hyperbilirubinemia, familial transient neonatal, 237900	191740	88	100	100	100
UMOD	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860	191845	62	100	99	85
UMPS	Orotic aciduria, 258900	613891	56	100	100	92
UNG	Immunodeficiency with hyper IgM, type 5, 608106	191525	60	100	100	99
UPB1	Beta-ureidopropionase deficiency, 613161	606673	65	100	100	96
UPF3B	Mental retardation, syndromic 14, 300676	300298	61	100	97	89
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158	191330	61	100	100	95
UQCRC2	Mitochondrial complex III deficiency, nuclear type 5, 615160	191329	67	100	99	84
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	86	100	100	100
UROC1	?Urocanase deficiency, 276880	613012	59	100	100	97
UROD	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100	613521	70	100	100	98
UROS	Porphyria, congenital erythropoietic, 263700	606938	45	100	99	81
USB1	Poikiloderma with neutropenia, 604173	613276	47	100	99	85
USH1C	Deafness 18A, 602092 Usher syndrome, type 1C, 276904	605242	57	100	98	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
USH1G	Usher syndrome, type 1G, 606943	607696	111	100	100	100
USH2A	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901	608400	59	100	99	92
USP21	No OMIM phenotype	604729	78	100	100	95
USP9Y	Spermatogenic failure, Y-linked, 2, 415000, Y-linked	400005	10	45	25	6
UTP4	No OMIM phenotype	607456	57	100	99	85
UVSSA	UV-sensitive syndrome 3, 614640	614632	77	100	100	99
VANGL1	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940	610132	74	100	100	94
VANGL2	Neural tube defects, 182940	600533	100	100	100	99
VAPB	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980	605704	50	100	100	85
VAX1	?Microphthalmia, syndromic 11, 614402	604294	59	99	92	86
VCAN	Wagner syndrome 1, 143200	118661	53	100	100	96
VCL	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255	193065	60	100	96	86
VCP	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320	601023	59	100	99	88
VDR	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440	601769	60	100	100	94
VHL	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300	608537	88	100	100	100
VIM	Cataract 30, pulverulent, 116300	193060	64	100	99	93
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404	613401	41	100	98	77
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700	608547	68	100	100	96
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	48	100	99	87
VPS13A	Choreoacanthocytosis, 200150	605978	55	100	98	84
VPS13B	Cohen syndrome, 216550	607817	54	100	98	87
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085	608552	48	100	97	87
VPS35	{Parkinson disease 17}, 614203	601501	52	100	97	85
VPS37A	Spastic paraplegia 53, 614898	609927	39	99	93	68
VPS45	Neutropenia, severe congenital, 5, 615285	610035	52	100	99	86
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	42	100	98	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
VSX1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300	605020	59	100	100	97
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	60	100	100	97
VWF	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480	613160	60	100	99	93
WAS	Neutropenia, severe congenital, 300299 Thrombocytopenia, 313900 Thrombocytopenia, intermittent, 313900 Wiskott-Aldrich syndrome, 301000	300392	54	96	84	75
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	45	100	97	78
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	53	100	98	84
WDR11	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858	606417	59	100	98	86
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	50	100	98	85
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	613363	72	100	100	97
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613602	43	100	97	77
WDR36	Glaucoma 1, open angle, G, 609887	609669	54	100	98	83
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	87	100	100	100
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	615462	56	100	98	85
WDR62	Microcephaly 2, primary, with or without cortical malformations, 604317	613583	79	100	100	98
WDR72	Amelogenesis imperfecta, type IIA3, 613211	613214	52	100	98	86
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185	614218	96	100	100	100
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome, 222300 Wolfram-like syndrome, 614296	606201	106	100	100	100
WHRN	Deafness 31, 607084 Usher syndrome, type 2D, 611383	607928	87	100	100	100
WIPF1	?Wiskott-Aldrich syndrome 2, 614493	602357	82	100	98	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230	603400	59	100	100	98
WNK1	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492	605232	63	100	100	97
WNK4	Pseudohypoaldosteronism, type IIB, 614491	601844	77	100	100	98
WNT1	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to}, 615221	164820	118	100	100	100
WNT10A	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400	606268	81	100	100	100
WNT10B	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073	601906	90	100	100	100
WNT3	?Tetra-amelia syndrome, 273395	165330	97	100	100	96
WNT4	Mullerian aplasia and hyperandrogenism, 158330 ?SERKAL syndrome, 611812	603490	115	92	92	92
WNT5A	Robinow syndrome 1, 180700	164975	76	100	100	91
WNT7A	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820	601570	87	100	100	100
WRAP53	Dyskeratosis congenita 3, 613988	612661	95	100	100	100
WRN	Werner syndrome, 277700	604611	47	100	98	84
WT1	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070	607102	80	100	98	96
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia 12, 614322	605131	57	100	100	93
XDH	Xanthinuria, type I, 278300	607633	53	100	98	85
XIAP	Lymphoproliferative syndrome, 2, 300635	300079	52	100	96	85
XK	McLeod syndrome with or without chronic granulomatous disease, 300842	314850	51	100	100	94
XPA	Xeroderma pigmentosum, group A, 278700	611153	45	100	92	70
XPC	Xeroderma pigmentosum, group C, 278720	613208	66	100	98	90
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	41	100	98	82
YAP1	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	49	100	95	79
YARS	Charcot-Marie-Tooth disease, dominant intermediate C, 608323	603623	62	100	100	93

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YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	610957	78	100	100	96
ZAP70	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840	176947	86	100	100	100
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	92	100	100	99
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069	614064	57	100	100	99
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	62	100	100	88
ZCCHC16	No OMIM phenotype	No id	54	100	100	100
ZDHHC9	Mental retardation syndromic, Raymond type, 300799	300646	39	100	95	69
ZEB1	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141	189909	62	100	100	97
ZEB2	Mowat-Wilson syndrome, 235730	605802	61	100	100	98
ZFP57	Diabetes mellitus, transient neonatal, 1, 601410	612192	74	100	100	98
ZFPM2	Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500 46XY sex reversal 9, 616067	603693	60	100	100	100
ZFYVE26	Spastic paraplegia 15, 270700	612012	64	100	99	91
ZFYVE27	Spastic paraplegia 33, 610244	610243	63	100	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	82	95	90	87
ZIC3	Congenital heart defects, nonsyndromic, 1, 306955 Heterotaxy, visceral, 1, 306955 VACTERL association, 314390	300265	60	100	97	86
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210	606480	42	100	97	77
ZMYND10	Ciliary dyskinesia, primary, 22, 615444	607070	92	100	100	100
ZNF335	?Microcephaly 10, primary, 615095	610827	78	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	121	100	100	99
ZNF469	Brittle cornea syndrome 1, 229200	612078	88	100	100	100
ZNF513	?Retinitis pigmentosa 58, 613617	613598	96	100	100	100
ZNF592	No OMIM phenotype	613624	87	100	100	100
ZNF644	Myopia 21, 614167	614159	54	100	100	99
ZNF711	Mental retardation 97, 300803	314990	50	100	93	83
ZNF750	Seborrhea-like dermatitis with psoriasiform elements, 610227	610226	121	100	100	100
ZNF81	No OMIM phenotype	314998	46	100	100	87

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x

- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 96 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x