Mendeliome

Gene panel

Gene panel information

Gene panel	Mendeliome
Version	7
Total genes	4969
Activation date	Thursday 24 october 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
A2ML1	99.97 %	610627	{Otitis media, susceptibility to}, 166760 (3), Autosomal dominant
A4GALT	100 %	607922	[Blood group, P1Pk system, P(2) phenotype], 111400 (3); NOR polyagglutination syndrome, 111400 (3); [Blood group, P1Pk system, p phenotype], 111400 (3)
AAAS	99.88 %	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AAGAB	100 %	614888	Keratoderma, palmoplantar, punctate type IA, 148600 (3), Autosomal dominant
AARS1	99.99 %	601065	Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive
AARS2	99.98 %	612035	Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive
AASS	99.61 %	605113	Hyperlysinemia, 238700 (3), Autosomal recessive
ABAT	99.98 %	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCA1	99.92 %	600046	Tangier disease, 205400 (3), Autosomal recessive; HDL deficiency, familial, 1, 604091 (3), Autosomal dominant
ABCA12	99.94 %	607800	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive
ABCA2	100 %	600047	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive
ABCA3	99.96 %	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
ABCA4	99.3 %	601691	Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3), Autosomal recessive; Fundus flavimaculatus, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive
ABCA5	99.72 %	612503	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400 (3), Autosomal recessive
ABCA7	99.97 %	605414	{Alzheimer disease 9, susceptibility to}, 608907 (3), Autosomal dominant
ABCB1	99.75 %	171050	{Inflammatory bowel disease 13}, 612244 (3); {Colchicine resistance}, 120080 (3)
ABCB11	99.86 %	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
ABCB4	99.71 %	171060	Gallbladder disease 1, 600803 (3), Autosomal dominant, Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3), Autosomal dominant, Autosomal recessive; Cholestasis, progressive familial intrahepatic 3, 602347 (3), Autosomal recessive

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ABCB6	99.97 %	605452	Microphthalmia, isolated, with coloboma 7, 614497 (3), Autosomal dominant; Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; [Blood group, Langereis system], 111600 (3); Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant
ABCB7	99.58 %	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked
ABCC1	99.34 %	158343	?Deafness, autosomal dominant 77, 618915 (3), Autosomal dominant
ABCC11	99.96 %	607040	[Axillary odor, variation in], 117800 (3), Autosomal dominant; [Earwax, wet/dry], 117800 (3), Autosomal dominant; [Colostrum secretion, variation in], 117800 (3), Autosomal dominant
ABCC2	99.94 %	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
ABCC6	98.57 %	603234	Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant
ABCC8	99.98 %	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal dominant, Autosomal recessive; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal dominant, Autosomal recessive
ABCC9	99.92 %	601439	Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
ABCD1	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABCD3	92.7 %	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
ABCD4	100 %	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
ABCG2	99.26 %	603756	[Junior blood group system], 614490 (3); [Uric acid concentration, serum, QTL1], 138900 (3), ?Autosomal dominant
ABCG5	99.96 %	605459	Sitosterolemia 2, 618666 (3), Autosomal recessive
ABCG8	99.95 %	605460	Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3)
ABHD12	99.98 %	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ABHD16A	100 %	142620	Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive
ABHD5	99.98 %	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ABL1	100 %	189980	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant
ABO	100 %	110300	[Blood group, ABO system], 616093 (3)
ACACA	99.92 %	200350	Acetyl-CoA carboxylase deficiency, 613933 (3), Autosomal recessive
ACAD8	99.97 %	604773	Isobutyryl-CoA dehydrogenase deficiency, 611283 (3), Autosomal recessive
ACAD9	100 %	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACADM	96.14 %	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
ACADS	99.99 %	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
ACADSB	99.93 %	600301	2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive

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ACADVL	100 %	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
ACAN	91.51 %	155760	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant; Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 (3), Autosomal recessive
ACAT1	99.81 %	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
ACAT2	99.99 %	100678	?ACAT2 deficiency, 614055 (1), Isolated cases
ACBD5	99.97 %	616618	Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive
ACBD6	98.86 %	616352	Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive
ACD	100 %	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal dominant, Autosomal recessive; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal dominant, Autosomal recessive
ACE	99.98 %	106180	{Stroke, hemorrhagic}, 614519 (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Myocardial infarction, susceptibility to} (3); {Microvascular complications of diabetes 3}, 612624 (3); [Angiotensin I-converting enzyme, benign serum increase] (3); {SARS, progression of} (3)
ACER3	99.76 %	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive
ACHE	99.99 %	100740	[Blood group, Yt system], 112100 (3)
ACKR1	99.98 %	613665	[Blood group, Duffy system], 110700 (3), Autosomal dominant, Autosomal recessive; [White blood cell count QTL], 611862 (3), Autosomal recessive; {Malaria, vivax, protection against}, 611162 (3)
ACKR3	100 %	610376	?Oculomotor-abducens synkinesis, 619215 (3), Autosomal recessive
ACO2	99.99 %	100850	Optic atrophy 9, 616289 (3), Autosomal dominant, Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
ACOX1	99.98 %	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACOX2	99.81 %	601641	Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive
ACP2	100 %	171650	?Lysosomal acid phosphatase deficiency, 200950 (1), Autosomal recessive
ACP4	99.93 %	606362	Amelogenesis imperfecta, type IJ, 617297 (3), Autosomal recessive
ACP5	100 %	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACR	97.24 %	102480	?Spermatogenic failure 87, 620500 (3), Autosomal recessive
ACSF3	99.99 %	614245	Combined malonic and methylmalonic aciduria, 614265 (3), Autosomal recessive
ACSL4	99.59 %	300157	Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant
ACSL5	99.96 %	605677	?Diarrhea 13, 620357 (3), Autosomal recessive
ACSL6	99.21 %	604443	Myelodysplastic syndrome (3); Myelogenous leukemia, acute (3)
ACSM3	99.47 %	145505	{?Hypertension, essential} (1)
ACTA1	99.99 %	102610	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265 (3), Autosomal recessive; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Congenital myopathy 2C, severe infantile, autosomal dominant, 620278 (3), Autosomal dominant; Congenital myopathy 2A, typical, autosomal dominant, 161800 (3), Autosomal dominant
ACTA2	99.99 %	102620	Smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant; Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3)

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ACTB	100 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479 (3)
ACTC1	98.57 %	102540	Left ventricular noncompaction 4, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant
ACTG1	100 %	102560	Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant
ACTG2	99.99 %	102545	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 (3), Autosomal dominant; Visceral myopathy 1, 155310 (3), Autosomal dominant
ACTL6B	99.9 %	612458	Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant
ACTL7A	100 %	604303	Spermatogenic failure 86, 620499 (3), Autosomal recessive
ACTL9	100 %	619251	Spermatogenic failure 53, 619258 (3), Autosomal recessive
ACTN1	100 %	102575	Bleeding disorder, platelet-type, 15, 615193 (3), Autosomal dominant
ACTN2	99.99 %	102573	Myopathy, distal, 6, adult onset, 618655 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant; Congenital myopathy 8, 618654 (3), Autosomal dominant; Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant
ACTN3	99.99 %	102574	[Sprinting performance], 617749 (3), Autosomal recessive; [Alpha-actinin-3 deficiency], 617749 (3), Autosomal recessive
ACTN4	100 %	604638	Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant
ACVR1	99.94 %	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACVR1B	99.99 %	601300	Pancreatic cancer, somatic, 260350 (3)
ACVR2B	99.99 %	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
ACVRL1	99.88 %	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
ACY1	100 %	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
ADA	99.97 %	608958	Adenosine deaminase deficiency, partial, 102700 (3), Somatic mosaicism, Autosomal recessive; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Somatic mosaicism, Autosomal recessive
ADA2	100 %	607575	Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive
ADAM10	99.88 %	602192	{Alzheimer disease 18, susceptibility to}, 615590 (3); Reticulate acropigmentation of Kitamura, 615537 (3), Autosomal dominant
ADAM17	99.94 %	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
ADAM22	99.15 %	603709	Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive
ADAM9	99.89 %	602713	Cone-rod dystrophy 9, 612775 (3), Autosomal recessive
ADAMTS10	99.99 %	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS13	100 %	604134	Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive
ADAMTS15	99.98 %	607509	Arthrogryposis, distal, type 12, 620545 (3), Autosomal recessive
ADAMTS17	99.99 %	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
ADAMTS18	99.99 %	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
ADAMTS19	99.94 %	607513	Cardiac valvular dysplasia 2, 620067 (3), Autosomal recessive

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ADAMTS2	97.58 %	604539	Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive
ADAMTS3	99.94 %	605011	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154 (3), Autosomal recessive
ADAMTSL2	99.99 %	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
ADAMTSL4	99.66 %	610113	Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive
ADAR	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADARB1	94.29 %	601218	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive
ADAT3	100 %	615302	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive
ADCY1	99.91 %	103072	?Deafness, autosomal recessive 44, 610154 (3), Autosomal recessive
ADCY10	99.77 %	605205	{Hypercalciuria, absorptive, susceptibility to}, 143870 (3), Autosomal dominant
ADCY3	99.94 %	600291	{Obesity, susceptibility to, BMIQ19}, 617885 (3), Autosomal recessive
ADCY5	99.98 %	600293	Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive
ADCY6	99.98 %	600294	Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive
ADD1	100 %	102680	{Hypertension, essential, salt-sensitive}, 145500 (3), Multifactorial
ADD3	99.95 %	601568	Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive
ADGRE2	98.77 %	606100	Vibratory urticaria, 125630 (3), Autosomal dominant
ADGRG1	99.9 %	604110	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752 (3); Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854 (3), Autosomal recessive
ADGRG2	99.69 %	300572	Congenital bilateral absence of vas deferens, X-linked, 300985 (3), X-linked
ADGRG6	99.91 %	612243	Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive
ADGRL1	99.98 %	616416	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065 (3), Autosomal dominant
ADGRV1	99.8 %	602851	Usher syndrome, type 2C, 605472 (3), Digenic dominant, Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant
ADH1B	99.97 %	103720	{Aerodigestive tract cancer, squamous cell, alcohol-related, protection against}, 103780 (3), Multifactorial; {Alcohol dependence, protection against}, 103780 (3), Multifactorial
ADH1C	99.94 %	103730	{Alcohol dependence, protection against}, 103780 (3), Multifactorial; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
ADH5	99.89 %	103710	AMED syndrome, digenic, 619151 (3), Digenic recessive
ADIPOQ	100 %	605441	Adiponectin deficiency, 612556 (3), Autosomal dominant
ADK	99.78 %	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
ADNP	100 %	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
ADPRS	99.94 %	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
ADRA2A	100 %	104210	?Lipodystrophy, familial partial, type 8, 620679 (3), Autosomal dominant

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ADRB1	100 %	109630	?[Short sleep, familial natural, 2], 618591 (3), Autosomal dominant; [Resting heart rate], 607276 (3)
ADRB2	100 %	109690	Beta-2-adrenoreceptor agonist, reduced response to (3)
ADRB3	100 %	109691	{Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive
ADSL	99.93 %	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
ADSS1	99.99 %	612498	Myopathy, distal, 5, 617030 (3), Autosomal recessive
AEBP1	99.97 %	602981	Ehlers-Danlos syndrome, classic-like, 2, 618000 (3), Autosomal recessive
AFF2	99.89 %	300806	Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive
AFF3	99.63 %	601464	KINSSHIP syndrome, 619297 (3), Autosomal dominant
AFF4	99.94 %	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AFG3L2	99.97 %	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AFP	99.46 %	104150	[Hereditary persistence of alpha-fetoprotein], 615970 (3), Autosomal dominant; Alpha-fetoprotein deficiency, 615969 (3), Autosomal recessive
AGA	99.92 %	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGBL1	99.99 %	615496	Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant
AGBL5	99.98 %	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
AGK	99.99 %	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
AGL	97.67 %	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
AGO1	99.73 %	606228	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292 (3), Autosomal dominant
AGO2	99.97 %	606229	Lessel-Kreienkamp syndrome, 619149 (3), Autosomal dominant
AGPAT2	100 %	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGPS	98.83 %	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
AGR2	99.98 %	606358	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233 (3), Autosomal recessive
AGRN	99.99 %	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive
AGRP	99.96 %	602311	{Leanness, inherited}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; {Obesity, late-onset}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive
AGT	100 %	106150	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Preeclampsia, susceptibility to} (3); {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
AGTPBP1	99.68 %	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
AGTR1	99.97 %	106165	{Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
AGXT	100 %	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AGXT2	100 %	612471	[Beta-aminoisobutyric acid, urinary excretion of], 210100 (3), Autosomal recessive
AHCY	100 %	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
AHDC1	100 %	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant

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AHI1	99.86 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AHR	99.79 %	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
AHSG	99.99 %	138680	?Alopecia-intellectual disability syndrome 1, 203650 (3), Autosomal recessive
AICDA	99.94 %	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
AIFM1	99.92 %	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
AIMP1	99.97 %	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
AIMP2	99.99 %	600859	Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive
AIP	99.99 %	605555	Pituitary adenoma 1, multiple types, 102200 (3), Somatic mutation, Autosomal dominant; Pituitary adenoma predisposition, 102200 (3), Somatic mutation, Autosomal dominant
AIPL1	100 %	604392	Leber congenital amaurosis 4, 604393 (3), Autosomal dominant, Autosomal recessive; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal dominant, Autosomal recessive; Cone-rod dystrophy, 604393 (3), Autosomal dominant, Autosomal recessive
AIRE	99.95 %	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal dominant, Autosomal recessive
AK1	100 %	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
AK2	99.39 %	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
AK7	99.93 %	615364	?Spermatogenic failure 27, 617965 (3), Autosomal recessive
AK9	99.53 %	615358	Spermatogenic failure 89, 620705 (3), Autosomal recessive
AKAP3	100 %	604689	Spermatogenic failure 82, 620353 (3), Autosomal recessive
AKAP9	99.27 %	604001	?Long QT syndrome 11, 611820 (3), Autosomal dominant
AKR1C2	91.19 %	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
AKR1C4	100 %	600451	{46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive
AKR1D1	99.91 %	604741	Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive
AKT1	100 %	164730	Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3); Colorectal cancer, somatic, 114500 (3); Proteus syndrome, somatic, 176920 (3); Ovarian cancer, somatic, 167000 (3)
AKT2	99.85 %	164731	Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant
AKT3	99.81 %	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant
ALAD	99.99 %	125270	Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive
ALAS2	99.98 %	301300	Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyria, erythropoietic, X-linked, 300752 (3), X-linked
ALB	99.71 %	103600	?[Dysalbuminemic hypertriiodothyroninemia], 615999 (3), Autosomal dominant, Autosomal recessive; Analbuminemia, 616000 (3), Autosomal recessive; [Dysalbuminemic hyperthyroxinemia], 615999 (3), Autosomal dominant, Autosomal recessive
ALDH18A1	99.96 %	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
ALDH1A2	99.97 %	603687	Diaphragmatic hernia 4, with cardiovascular defects, 620025 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ALDH1A3	99.96 %	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
ALDH2	100 %	100650	{Esophageal cancer, alcohol-related, susceptibility to} (3); {Sublingual nitroglycerin, susceptibility to poor response to} (3); Alcohol sensitivity, acute, 610251 (3), Autosomal dominant; {Hangover, susceptibility to}, 610251 (3), Autosomal dominant
ALDH3A2	99.95 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH4A1	98.97 %	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
ALDH5A1	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALDH6A1	99.99 %	603178	Methylmalonate semialdehyde dehydrogenase deficiency, 614105 (3), Autosomal recessive
ALDH7A1	99.49 %	107323	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100 (3), Autosomal recessive
ALDOA	100 %	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive
ALDOB	100 %	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
ALG1	86.66 %	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG10B	99.97 %	603313	{Long QT syndrome, acquired, reduced susceptibility to}, 613688 (3), Autosomal dominant
ALG11	99.99 %	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
ALG12	100 %	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
ALG13	99.44 %	300776	Developmental and epileptic encephalopathy 36, 300884 (3), X-linked
ALG14	99.34 %	612866	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3), Autosomal recessive; Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive
ALG2	100 %	607905	Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
ALG3	99.96 %	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
ALG5	99.94 %	604565	Polycystic kidney disease 7, 620056 (3), Autosomal dominant
ALG6	93.37 %	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
ALG8	95.49 %	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
ALG9	99.73 %	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
ALK	99.93 %	105590	{Neuroblastoma, susceptibility to, 3}, 613014 (3)
ALKBH8	99.98 %	613306	Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive
ALMS1	99.9 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALOX12B	100 %	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
ALOX5	99.99 %	152390	{Atherosclerosis, susceptibility to} (3); {Asthma, diminished response to antileukotriene treatment in}, 600807 (3), Autosomal dominant
ALOX5AP	99.99 %	603700	{Stroke, susceptibility to}, 601367 (3), Multifactorial
ALOXE3	99.98 %	607206	Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive
ALPK1	99.92 %	607347	ROSAH syndrome, 614979 (3), Autosomal dominant
ALPK3	99.99 %	617608	Cardiomyopathy, familial hypertrophic 27, 618052 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ALPL	99.88 %	171760	Odontohypophosphatasia, 146300 (3), Autosomal dominant, Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal dominant, Autosomal recessive
ALS2	99.87 %	606352	Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive
ALX1	97.99 %	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
ALX3	99.74 %	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
ALX4	100 %	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
AMACR	100 %	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
AMBN	99.74 %	601259	Amelogenesis imperfecta, type IF, 616270 (3), Autosomal recessive
AMELX	99.98 %	300391	Amelogenesis imperfecta, type 1E, 301200 (3), X-linked dominant
AMER1	100 %	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
AMFR	99.74 %	603243	Spastic paraplegia 89, autosomal recessive, 620379 (3), Autosomal recessive
AMH	100 %	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
AMHR2	99.96 %	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
AMMECR1	99.87 %	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
AMN	100 %	605799	Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive
AMPD1	98.94 %	102770	Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive
AMPD2	99.91 %	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, autosomal recessive, 615686 (3), Autosomal recessive
AMPD3	99.99 %	102772	[AMP deaminase deficiency, erythrocytic], 612874 (3), Autosomal recessive
AMT	100 %	238310	Glycine encephalopathy 2, 620398 (3)
AMTN	99.95 %	610912	?Amelogenesis imperfecta, type IIIB, 617607 (3), Autosomal dominant
ANAPC1	75.83 %	608473	Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive
ANAPC7	99.99 %	606949	Ferguson-Bonni neurodevelopmental syndrome, 619699 (3), Autosomal recessive
ANG	100 %	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
ANGPT1	99.97 %	601667	?Angioedema, hereditary, 5, 619361 (3), Autosomal dominant
ANGPT2	99.94 %	601922	Lymphatic malformation 10, 619369 (3), Autosomal dominant
ANGPTL3	97.46 %	604774	Hypobetalipoproteinemia, familial, 2, 605019 (3), Autosomal recessive
ANGPTL4	100 %	605910	Plasma triglyceride level QTL, low, 615881 (3), Autosomal dominant
ANK1	99.98 %	612641	Spherocytosis, type 1, 182900 (3), Autosomal dominant, Autosomal recessive
ANK2	99.98 %	106410	Long QT syndrome 4, 600919 (3), Autosomal dominant; Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant
ANK3	99.79 %	600465	Intellectual developmental disorder, autosomal recessive 37, 615493 (3), Autosomal recessive
ANKH	99.95 %	605145	Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant
ANKLE2	99.99 %	616062	Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
ANKRD11	99.85 %	611192	KBG syndrome, 148050 (3), Autosomal dominant
ANKRD17	99.92 %	615929	Chopra-Amiel-Gordon syndrome, 619504 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ANKRD26	97.72 %	610855	Thrombocytopenia 2, 188000 (3), Autosomal dominant
ANKS6	100 %	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
ANLN	99.85 %	616027	Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant
ANO1	99.99 %	610108	Moyamoya disease 7, 620687 (3), Autosomal dominant, Autosomal recessive; ?Intestinal dysmotility syndrome, 620045 (3), Autosomal recessive
ANO10	99.93 %	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
ANO3	99.98 %	610110	Dystonia 24, 615034 (3), Autosomal dominant
ANO5	99.85 %	608662	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
ANO6	98.43 %	608663	Scott syndrome, 262890 (3), Autosomal recessive
ANOS1	99.96 %	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
ANTXR1	99.96 %	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
ANTXR2	99.67 %	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
ANXA11	99.71 %	602572	Amyotrophic lateral sclerosis 23, 617839 (3), Autosomal dominant; Inclusion body myopathy and brain white matter abnormalities, 619733 (3), Autosomal dominant
ANXA5	99.91 %	131230	{Pregnancy loss, recurrent, susceptibility to, 3}, 614391 (3), Autosomal dominant
AOPEP	99.99 %	619600	Dystonia 31, 619565 (3), Autosomal recessive
AP1B1	99.99 %	600157	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive
AP1G1	99.95 %	603533	Usmani-Riazuddin syndrome, autosomal recessive, 619548 (3), Autosomal recessive; Usmani-Riazuddin syndrome, autosomal dominant, 619467 (3), Autosomal dominant
AP1S1	99.49 %	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP1S2	99.56 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
AP1S3	100 %	615781	{Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant
AP2M1	99.69 %	601024	Intellectual developmental disorder 60 with seizures, 618587 (3), Autosomal dominant
AP2S1	99.98 %	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant
AP3B1	99.89 %	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3B2	100 %	602166	Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive
AP3D1	100 %	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
AP4B1	96.92 %	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
AP4E1	99.94 %	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
AP4M1	99.98 %	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
AP4S1	87.89 %	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
AP5Z1	100 %	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
APC	99.97 %	611731	Colorectal cancer, somatic, 114500 (3); Brain tumor-polyposis syndrome 2, 175100 (3), Autosomal dominant; Desmoid disease, hereditary, 135290 (3), Autosomal dominant; Adenoma, periampullary, somatic, 175100 (3); Hepatoblastoma, somatic, 114550 (3); Gastric cancer, somatic, 613659 (3); Gastric adenocarcinoma and proximal polyposis of the stomach, 619182 (3), Autosomal dominant; Gardner syndrome, 175100 (3), Autosomal dominant; Adenomatous polyposis coli, 175100 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
APC2	99.98 %	612034	Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 74, 617169 (3), Autosomal recessive
APCDD1	99.99 %	607479	Hypotrichosis 1, 605389 (3), Autosomal dominant
APOA1	100 %	107680	Hypoalphalipoproteinemia, primary, 2, 618463 (3), Autosomal recessive; Amyloidosis, hereditary systemic 3, 620657 (3); Hypoalphalipoproteinemia, primary, 2, intermediate, 619836 (3), Autosomal dominant
APOA2	99.84 %	107670	Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant, Autosomal recessive
APOA5	100 %	606368	Hyperchylomicronemia, late-onset, 144650 (3), Autosomal dominant; {Hypertriglyceridemia, susceptibility to}, 145750 (3), Autosomal dominant
APOB	99.99 %	107730	Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive
APOC2	99.97 %	608083	Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive
APOC3	100 %	107720	Apolipoprotein C-III deficiency, 614028 (3)
APOE	99.98 %	107741	Alzheimer disease 2, 104310 (3), Autosomal dominant; Sea-blue histiocyte disease, 269600 (3), Autosomal recessive; {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 (3), Autosomal dominant; {Coronary artery disease, severe, susceptibility to}, 617347 (3); Lipoprotein glomerulopathy, 611771 (3); {?Macular degeneration, age-related}, 603075 (3), Autosomal dominant; Hyperlipoproteinemia, type III, 617347 (3)
APOL1	99.99 %	603743	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3), Autosomal dominant
APOL2	99.99 %	607252	{Schizophrenia}, 181500 (1), Autosomal dominant
APOL4	99.96 %	607254	{Schizophrenia}, 181500 (1), Autosomal dominant
APOLD1	100 %	612456	?Bleeding disorder, vascular-type, 620715 (3), Autosomal dominant
APP	99.92 %	104760	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant; Alzheimer disease 1, familial, 104300 (3), Autosomal dominant
APPL1	99.69 %	604299	{Maturity-onset diabetes of the young, type 14}, 616511 (3), Autosomal dominant
APRT	100 %	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
APTX	99.92 %	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
AQP1	99.99 %	107776	[Aquaporin-1 deficiency], 110450 (3); [Blood group, Colton], 110450 (3)
AQP2	100 %	107777	Diabetes insipidus, nephrogenic, 2, 125800 (3), Autosomal dominant, Autosomal recessive
AQP3	99.95 %	600170	[Blood group GIL], 607457 (3)
AQP4	100 %	600308	?Megalencephalic leukoencephalopathy with subcortical cysts 4, remitting, 620448 (3), Autosomal recessive
AQP5	99.88 %	600442	Palmoplantar keratoderma, Bothnian type, 600231 (3), Autosomal dominant
AQP7	100 %	602974	[Glycerol quantitative trait locus], 614411 (3), Autosomal recessive
AR	99.76 %	313700	Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Spinal and bulbar muscular atrophy, X-linked 1, 313200 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 301120 (3), X-linked; Androgen insensitivity, 300068 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive
ARCN1	99.92 %	600820	Short stature-micrognathia syndrome, 617164 (3), Autosomal dominant
ARF1	99.99 %	103180	Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ARFGEF1	99.86 %	604141	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964 (3), Autosomal dominant
ARFGEF2	99.99 %	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
ARG1	99.95 %	608313	Argininemia, 207800 (3), Autosomal recessive
ARHGAP26	99.99 %	605370	Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
ARHGAP29	93.54 %	610496	*No OMIM phenotypes*
ARHGAP31	100 %	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARHGDIA	100 %	601925	Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive
ARHGEF1	99.97 %	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
ARHGEF10	100 %	608136	?Slowed nerve conduction velocity, AD, 608236 (3), Autosomal dominant
ARHGEF18	99.97 %	616432	Retinitis pigmentosa 78, 617433 (3), Autosomal recessive
ARHGEF2	99.91 %	607560	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523 (3), Autosomal recessive
ARHGEF9	99.89 %	300429	Developmental and epileptic encephalopathy 8, 300607 (3), X-linked
ARID1A	99.83 %	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	99.69 %	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARID2	99.48 %	609539	Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant
ARL13B	99.53 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL2	99.84 %	601175	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082 (3), Autosomal dominant
ARL2BP	99.89 %	615407	Retinitis pigmentosa 82 with or without situs inversus, 615434 (3), Autosomal recessive
ARL3	99.98 %	604695	Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive
ARL6	99.9 %	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
ARL6IP1	99.51 %	607669	Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive
ARMC12	100 %	620377	Spermatogenic failure 90, 620744 (3), Autosomal recessive
ARMC2	99.97 %	618424	Spermatogenic failure 38, 618433 (3), Autosomal recessive
ARMC5	99.97 %	615549	ACTH-independent macronodular adrenal hyperplasia 2, 615954 (3), Somatic mutation, Autosomal dominant
ARMC9	99.77 %	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
ARMS2	100 %	611313	{Macular degeneration, age-related, 8}, 613778 (3)
ARNT2	99.96 %	606036	?Webb-Dattani syndrome, 615926 (3), Autosomal recessive
ARPC1B	99.92 %	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive
ARPC4	100 %	604226	Developmental delay, language impairment, and ocular abnormalities, 620141 (3), Autosomal dominant
ARPC5	99.85 %	604227	Immunodeficiency 133 with autoimmunity and autoinflammation, 620565 (3), Autosomal recessive
ARR3	99.94 %	301770	Myopia 26, X-linked, female-limited, 301010 (3), X-linked
ARSA	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSB	99.99 %	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
ARSG	100 %	610008	Usher syndrome, type IV, 618144 (3), Autosomal recessive
ARSK	99.91 %	610011	Mucopolysaccharidosis, type X, 619698 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ARSL	99.93 %	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ART4	99.87 %	110600	[Blood group, Dombrock], 616060 (3)
ARV1	99.85 %	611647	Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive
ARX	95.36 %	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
ASAH1	99.9 %	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASB10	100 %	615054	Glaucoma 1, open angle, F, 603383 (3), Autosomal dominant
ASCC1	90.99 %	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
ASCC3	99.76 %	614217	Intellectual developmental disorder, autosomal recessive 81, 620700 (3), Autosomal recessive
ASH1L	99.79 %	607999	Intellectual developmental disorder, autosomal dominant 52, 617796 (3), Autosomal dominant
ASIP	100 %	600201	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 (3); [Skin/hair/eye pigmentation 9, dark/light hair], 611742 (3)
ASL	99.98 %	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASNS	99.63 %	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
ASPA	99.98 %	608034	Canavan disease, 271900 (3), Autosomal recessive
ASPH	99.92 %	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
ASPM	99.57 %	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
ASPN	99.96 %	608135	{Lumbar disc degeneration}, 603932 (3); {Osteoarthritis susceptibility 3}, 607850 (3), Autosomal dominant
ASPRV1	100 %	611765	Ichthyosis, lamellar, autosomal dominant, 146750 (3), Autosomal dominant
ASPSCR1	100 %	606236	Alveolar soft-part sarcoma, 606243 (3)
ASS1	77.52 %	603470	Citrullinemia, 215700 (3), Autosomal recessive
ASTL	99.39 %	608860	?Oocyte/zygote/embryo maturation arrest 11, 619643 (3), Autosomal recessive
ASXL1	100 %	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
ASXL2	99.82 %	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ASXL3	99.99 %	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant
ATAD1	99.85 %	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
ATAD3A	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATCAY	100 %	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATF6	98.19 %	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
ATG16L1	99.95 %	610767	{Inflammatory bowel disease (Crohn disease) 10}, 611081 (3)
ATG5	99.81 %	604261	?Spinocerebellar ataxia, autosomal recessive 25, 617584 (3), Autosomal recessive
ATG7	99.9 %	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive
ATIC	99.86 %	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATL1	99.95 %	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
ATL3	99.75 %	609369	Neuropathy, hereditary sensory, type IF, 615632 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ATM	99.83 %	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATN1	99.9 %	607462	Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant
ATOH1	99.99 %	601461	?Deafness, autosomal dominant 89, 620284 (3), Autosomal dominant
ATOH7	100 %	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
ATP11A	99.81 %	605868	?Auditory neuropathy, autosomal dominant 2, 620384 (3), Autosomal dominant; ?Leukodystrophy, hypomyelinating, 24, 619851 (3), Autosomal dominant; Deafness, autosomal dominant 84, 619810 (3), Autosomal dominant
ATP11C	99.66 %	300516	?Hemolytic anemia, congenital, X-linked, 301015 (3), X-linked recessive
ATP13A2	99.96 %	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
ATP13A3	99.84 %	610232	Pulmonary hypertension, primary, 5, 265400 (3), Autosomal recessive
ATP1A1	98.41 %	182310	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	99.98 %	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
ATP1B1	99.88 %	182330	[Blood pressure regulation QTL], 145500 (2), Multifactorial
ATP2A1	99.83 %	108730	Brody myopathy, 601003 (3), Autosomal recessive
ATP2A2	99.98 %	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP2B1	99.39 %	108731	Intellectual developmental disorder, autosomal dominant 66, 619910 (3), Autosomal dominant
ATP2B2	99.99 %	108733	Deafness, autosomal dominant 82, 619804 (3), Autosomal dominant; {Deafness, autosomal recessive 12, modifier of}, 601386 (3), Autosomal recessive
ATP2B3	99.98 %	300014	?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive
ATP2C1	100 %	604384	Hailey-Hailey disease, 169600 (3), Autosomal dominant
ATP5F1A	99.99 %	164360	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358 (3), Autosomal dominant; ?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive; ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228 (3), Autosomal recessive
ATP5F1B	99.79 %	102910	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085 (3), Autosomal dominant
ATP5F1D	99.99 %	603150	Mitochondrial complex V (ATP synthase) deficiency, 618120 (3), Autosomal recessive
ATP5F1E	100 %	606153	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ATP5MC3	99.97 %	602736	Dystonia, early-onset, and/or spastic paraplegia, 619681 (3), Autosomal dominant
ATP5MK	99.76 %	615204	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683 (3), Autosomal recessive
ATP5PO	99.94 %	600828	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 (3), Autosomal recessive
ATP6AP1	100 %	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
ATP6AP2	99.55 %	300556	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive
ATP6V0A1	99.85 %	192130	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant
ATP6V0A2	99.92 %	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
ATP6V0A4	99.93 %	605239	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722 (3), Autosomal recessive
ATP6V0C	100 %	108745	Epilepsy, early-onset, 3, with or without developmental delay, 620465 (3), Autosomal dominant
ATP6V1A	99.73 %	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant
ATP6V1B1	99.98 %	192132	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300 (3), Autosomal recessive
ATP6V1B2	99.99 %	606939	Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant
ATP6V1E1	99.99 %	108746	Cutis laxa, autosomal recessive, type IIC, 617402 (3), Autosomal recessive
ATP7A	99.87 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	100 %	605870	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATP8B1	99.94 %	602397	Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive
ATP9A	99.99 %	609126	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242 (3), Autosomal recessive
ATPAF2	99.96 %	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
ATR	99.83 %	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
ATRX	99.44 %	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive; Alpha-thalassemia/impaired intellectual development syndrome, 301040 (3), X-linked dominant
ATXN1	100 %	601556	Spinocerebellar ataxia 1, 164400 (3), Autosomal dominant
ATXN10	99.97 %	611150	Spinocerebellar ataxia 10, 603516 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ATXN2	99.88 %	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
ATXN3	99.99 %	607047	{Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Machado-Joseph disease, 109150 (3), Autosomal dominant
ATXN7	99.98 %	607640	Spinocerebellar ataxia 7, 164500 (3), Autosomal dominant
AUH	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
AURKA	100 %	603072	{Colon cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant
AURKC	99.57 %	603495	Spermatogenic failure 5, 243060 (3), Autosomal recessive
AUTS2	99.87 %	607270	Intellectual developmental disorder, autosomal dominant 26, 615834 (3), Autosomal dominant
AVIL	99.98 %	613397	Nephrotic syndrome, type 21, 618594 (3), Autosomal recessive
AVP	100 %	192340	Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant
AVPR2	100 %	300538	Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
AXIN1	100 %	603816	Hepatocellular carcinoma, somatic, 114550 (3); Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558 (3), Autosomal recessive; ?Caudal duplication anomaly, 607864 (3)
AXIN2	100 %	604025	Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant
B2M	100 %	109700	Amyloidosis, hereditary systemic 6, 620659 (3); Immunodeficiency 43, 241600 (3), Autosomal recessive
B3GALNT1	100 %	603094	[Blood group, P1PK system, P(k) phenotype], 111400 (3); [Blood group, globoside system], 615021 (3)
B3GALNT2	92.79 %	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive
B3GALT6	100 %	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive
B3GAT3	87.74 %	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
B3GLCT	99.9 %	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALNT1	99.97 %	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
B4GALNT2	99.87 %	111730	[Blood group, Sid system], 615018 (3); Sd(a) polyagglutination syndrome, 615018 (3)
B4GALT1	99.97 %	137060	Combined low LDL and fibrinogen, 620364 (3), Autosomal recessive; Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive
B4GALT7	99.99 %	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
B4GAT1	100 %	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
B9D1	99.8 %	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	99.88 %	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
BAAT	99.98 %	602938	Bile acid conjugation defect 1, 619232 (3), Autosomal recessive
BACH2	99.99 %	605394	Immunodeficiency 60 and autoimmunity, 618394 (3), Autosomal dominant
BAG3	100 %	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
BAG5	100 %	603885	Cardiomyopathy, dilated, 2F, 619747 (3), Autosomal recessive
BANF1	99.84 %	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
BAP1	99.99 %	603089	Kury-Isidor syndrome, 619762 (3), Autosomal dominant; Tumor predisposition syndrome 1, 614327 (3), Autosomal dominant; {Uveal melanoma, susceptibility to, 2}, 606661 (3), Autosomal dominant
BARD1	99.89 %	601593	{Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant
BAX	100 %	600040	Colorectal cancer, somatic, 114500 (3); T-cell acute lymphoblastic leukemia, somatic, 613065 (3)
BBIP1	99.99 %	613605	Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	99.98 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	100 %	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	99.9 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BBS4	99.88 %	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	99 %	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	99.42 %	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	99.75 %	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCAM	99.99 %	612773	[Blood group, Lutheran system], 111200 (3); [Blood group, Auberger system], 111200 (3); [Blood group, Lutheran null], 247420 (3), Autosomal recessive
BCAP31	99.95 %	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCAS3	99.31 %	607470	Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive
BCAT2	100 %	113530	Hypervalinemia or hyperleucine-isoleucinemia, 618850 (3), Autosomal recessive
BCHE	99.98 %	177400	Butyrylcholinesterase deficiency, 617936 (3), Autosomal recessive; {Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936 (3), Autosomal recessive
BCKDHA	99.97 %	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	99.73 %	248611	Maple syrup urine disease, type Ib, 620698 (3), Autosomal recessive
BCKDK	99.99 %	614901	Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3), Autosomal recessive
BCL10	99.74 %	603517	{Lymphoma, follicular, somatic}, 605027 (3); ?Immunodeficiency 37, 616098 (3), Autosomal recessive; {Sezary syndrome, somatic} (3); {Male germ cell tumor, somatic}, 273300 (3); Lymphoma, MALT, somatic, 137245 (3); {Mesothelioma, somatic}, 156240 (3)
BCL11A	99.93 %	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant
BCL11B	100 %	606558	Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant
BCL2	100 %	151430	Leukemia/lymphoma, B-cell, 2 (3)
BCL3	99.98 %	109560	Leukemia/lymphoma, B-cell, 3, 109560 (2)
BCO1	99.99 %	605748	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 (3), Autosomal dominant
BCOR	99.97 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BCORL1	99.99 %	300688	Shukla-Vernon syndrome, 301029 (3), X-linked recessive
BCR	99.79 %	151410	Leukemia, chronic myeloid, Philadelphia chromosome positive, somatic, 608232 (4); Leukemia, acute lymphocytic, Philadelphia chromosome positive, somatic, 613065 (4)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
BCS1L	99.99 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
BDP1	99.9 %	607012	?Deafness, autosomal recessive 112, 618257 (3), Autosomal recessive
BEAN1	95.6 %	612051	Spinocerebellar ataxia 31, 117210 (3), Autosomal dominant
BEST1	99.86 %	607854	Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 (3), Autosomal dominant; Retinitis pigmentosa-50, 613194 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3)
BET1	99.38 %	605456	Muscular dystrophy, congenital, with rapid progression, 254100 (3), Autosomal recessive
BFSP1	100 %	603307	Cataract 33, multiple types, 611391 (3), Autosomal dominant, Autosomal recessive
BFSP2	99.09 %	603212	Cataract 12, multiple types, 611597 (3), Autosomal dominant
BGN	99.97 %	301870	Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive
BHLHA9	100 %	615416	?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive; Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive
BHLHE41	99.6 %	606200	[Short sleep, familial natural, 1], 612975 (3), Autosomal dominant
BICC1	99.76 %	614295	{Renal dysplasia, cystic, susceptibility to}, 601331 (3), Autosomal dominant
BICD2	99.99 %	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
BICRA	99.99 %	605690	Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant
BIN1	99.98 %	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive
BLK	99.98 %	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
BLM	99.8 %	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLNK	99.9 %	604515	?Agammaglobulinemia 4, 613502 (3), Autosomal recessive
BLOC1S3	100 %	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S5	99.8 %	607289	Hermansky-Pudlak syndrome 11, 619172 (3), Autosomal recessive
BLOC1S6	99.98 %	604310	Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
BLVRA	99.95 %	109750	Hyperbiliverdinemia, 614156 (3), Autosomal dominant, Autosomal recessive
BMP1	99.94 %	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
BMP15	99.98 %	300247	Premature ovarian failure 4, 300510 (3), X-linked; Ovarian dysgenesis 2, 300510 (3), X-linked
BMP2	99.79 %	112261	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive
BMP4	100 %	112262	Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BMP6	100 %	112266	{Iron overload, susceptibility to}, 620121 (3), Autosomal dominant
BMPER	99.94 %	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
BMPR1A	99.58 %	601299	Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
BMPR1B	99.61 %	603248	Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant
BMPR2	99.95 %	600799	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3), Autosomal dominant; Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3), Autosomal dominant; Pulmonary venoocclusive disease 1, 265450 (3), Autosomal dominant
BMS1	85.8 %	611448	?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant
BNC1	100 %	601930	?Premature ovarian failure 16, 618723 (3), Autosomal dominant
BNC2	99.97 %	608669	Lower urinary tract obstruction, congenital, 618612 (3), Autosomal dominant
BOLA3	99.22 %	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
BPGM	100 %	613896	Erythrocytosis, familial, 8, 222800 (3), Autosomal recessive
BPNT2	100 %	614010	Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive
BPTF	99.84 %	601819	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant; {Kaposi sarcoma, susceptibility to}, 148000 (3), Autosomal dominant
BRAF	99.78 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
BRAT1	100 %	614506	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
BRCA1	98.33 %	113705	Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Breast-ovarian cancer, familial, 1}, 604370 (3), Autosomal dominant, Multifactorial; {Pancreatic cancer, susceptibility to, 4}, 614320 (3)
BRCA2	99.99 %	600185	Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal dominant, Autosomal recessive; {Prostate cancer}, 176807 (3), Somatic mutation, Autosomal dominant; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; {Breast cancer, male, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Pancreatic cancer 2}, 613347 (3); Wilms tumor, 194070 (3), Somatic mutation, Autosomal dominant
BRD4	99.98 %	608749	Cornelia de Lange syndrome 6, 620568 (3), Autosomal dominant
BRDT	93.71 %	602144	?Spermatogenic failure 21, 617644 (3), Autosomal recessive
BRF1	100 %	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
BRIP1	99.39 %	605882	Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant
BRPF1	100 %	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
BRWD1	99.88 %	617824	Ciliary dyskinesia, primary, 51, 620438 (3), Autosomal recessive
BRWD3	99.4 %	300553	Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
BSCL2	99.99 %	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
BSG	100 %	109480	[Blood group, OK], 111380 (3)
BSND	99.92 %	606412	Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive
BTD	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
BTG4	99.98 %	605673	Oocyte/zygote/embryo maturation arrest 8, 619009 (3), Autosomal recessive
BTK	99.88 %	300300	Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive
BTNL2	99.99 %	606000	{Sarcoidosis, susceptibility to, 2}, 612387 (3), Autosomal dominant
BUB1	99.64 %	602452	Colorectal cancer with chromosomal instability, somatic, 114500 (3); Microcephaly 30, primary, autosomal recessive, 620183 (3), Autosomal recessive
BUB1B	100 %	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
BVES	99.83 %	604577	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive
C11orf80	99.89 %	616109	Hydatidiform mole, recurrent, 4, 618432 (3), Autosomal recessive
C12orf4	99.93 %	616082	Intellectual developmental disorder, autosomal recessive 66, 618221 (3), Autosomal recessive
C12orf57	100 %	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
C14orf39	99.76 %	617307	Spermatogenic failure 52, 619202 (3), Autosomal recessive; ?Premature ovarian failure 18, 619203 (3), Autosomal recessive
C18orf32	100 %	619979	?Glycosylphosphatidylinositol biosynthesis defect 25, 619985 (3), Autosomal recessive
C19orf12	99.99 %	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
C1GALT1C1	99.95 %	300611	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110 (3), X-linked recessive; Tn polyagglutination syndrome, somatic, 300622 (3)
C1QA	99.99 %	120550	C1q deficiency 1, 613652 (3), Autosomal recessive
C1QB	99.58 %	120570	C1q deficiency 2, 620321 (3), Autosomal recessive
C1QBP	99.92 %	601269	Combined oxidative phosphorylation deficiency 33, 617713 (3), Autosomal recessive
C1QC	99.97 %	120575	C1q deficiency 3, 620322 (3), Autosomal recessive
C1QTNF5	100 %	608752	Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant
C1R	99.99 %	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
C1S	99.98 %	120580	C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant
C2	99.99 %	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
C2CD3	99.88 %	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
C2CD6	99.29 %	619776	?Spermatogenic failure 68, 619805 (3), Autosomal recessive
C2orf69	99.97 %	619219	Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
C3	100 %	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
C3orf52	99.8 %	611956	Hypotrichosis 15, 620177 (3), Autosomal recessive
C4A	18.13 %	120810	[Blood group, Rodgers], 614374 (3); C4a deficiency, 614380 (3), Autosomal recessive
C4B	21.28 %	120820	C4B deficiency, 614379 (3)
C5	99.92 %	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant
C6	99.97 %	217050	C6 deficiency, 612446 (3), Autosomal recessive
C7	99.94 %	217070	C7 deficiency, 610102 (3)
C8A	99.95 %	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
C8B	99.37 %	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
C9	99.89 %	120940	C9 deficiency, 613825 (3); {Macular degeneration, age-related, 15, susceptibility to}, 615591 (3), Autosomal dominant
C9orf72	99.9 %	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant
CA12	99.98 %	603263	Hyperchlorhidrosis, isolated, 143860 (3), Autosomal recessive
CA2	99.62 %	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CA5A	99.99 %	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
CA8	99.71 %	114815	Spinocerebellar ataxia, autosomal recessive 34, 613227 (3), Autosomal recessive
CABP2	99.98 %	607314	Deafness, autosomal recessive 93, 614899 (3), Autosomal recessive
CABP4	100 %	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
CACNA1A	98.16 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1B	100 %	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive
CACNA1C	100 %	114205	Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3), Autosomal dominant
CACNA1D	99.98 %	114206	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive
CACNA1E	99.82 %	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
CACNA1F	99.94 %	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
CACNA1G	99.95 %	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CACNA1H	100 %	607904	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3)
CACNA1I	99.98 %	608230	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114 (3), Autosomal dominant
CACNA1S	99.96 %	114208	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; Congenital myopathy 18 due to dihydropyridine receptor defect, 620246 (3), Autosomal dominant, Autosomal recessive; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant
CACNA2D1	97.12 %	114204	Developmental and epileptic encephalopathy 110, 620149 (3), Autosomal recessive
CACNA2D2	99.99 %	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
CACNA2D4	99.99 %	608171	Retinal cone dystrophy 4, 610478 (3), Autosomal recessive
CACNB2	99.93 %	600003	Brugada syndrome 4, 611876 (3), Autosomal dominant
CACNB4	99.2 %	601949	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; ?Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant
CACNG2	99.99 %	602911	?Intellectual developmental disorder, autosomal dominant 10, 614256 (3), Autosomal dominant
CAD	99.86 %	114010	Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive
CADM3	99.61 %	609743	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519 (3), Autosomal dominant
CALCR	99.19 %	114131	{Osteoporosis, postmenopausal, susceptibility}, 166710 (3), Autosomal dominant
CALCRL	99.53 %	114190	?Lymphatic malformation 8, 618773 (3), Autosomal recessive
CALM1	99.86 %	114180	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3), Autosomal dominant; Long QT syndrome 14, 616247 (3), Autosomal dominant
CALM2	99.62 %	114182	Long QT syndrome 15, 616249 (3), Autosomal dominant
CALM3	100 %	114183	Long QT syndrome 16, 618782 (3), Autosomal dominant; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782 (3), Autosomal dominant
CALR	100 %	109091	Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3)
CAMK2A	99.99 %	114078	Intellectual developmental disorder, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 63, 618095 (3), Autosomal recessive
CAMK2B	99.92 %	607707	Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant
CAMK2G	99.95 %	602123	Intellectual developmental disorder, autosomal dominant 59, 618522 (3), Autosomal dominant
CAMLG	97.9 %	601118	?Congenital disorder of glycosylation, type IIz, 620201 (3), Autosomal recessive
CAMSAP1	99.99 %	613774	Cortical dysplasia, complex, with other brain malformations 12, 620316 (3), Autosomal recessive
CAMTA1	99.97 %	611501	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant
CANT1	100 %	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive
CAP2	99.99 %	618385	Cardiomyopathy, dilated, 2I, 620462 (3), Autosomal recessive
CAPN1	99.99 %	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive
CAPN10	99.99 %	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CAPN15	99.98 %	603267	Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive
CAPN3	99.99 %	114240	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant
CAPN5	99.88 %	602537	Vitreoretinopathy, neovascular inflammatory, 193235 (3), Autosomal dominant
CAPNS1	99.96 %	114170	Pulmonary hypertension, primary, 6, 620777 (3), Autosomal recessive
CAPRIN1	99.56 %	601178	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782 (3), Autosomal dominant; Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636 (3), Autosomal dominant
CARD10	99.99 %	607209	?Immunodeficiency 89 and autoimmunity, 619632 (3), Autosomal recessive
CARD11	99.97 %	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive
CARD14	99.99 %	607211	Psoriasis 2, 602723 (3), Autosomal dominant; Pityriasis rubra pilaris, 173200 (3), Autosomal dominant
CARD8	99.99 %	609051	?Inflammatory bowel disease (Crohn disease) 30, 619079 (3), Autosomal dominant
CARD9	100 %	607212	Immunodeficiency 103, susceptibility to fungal infection, 212050 (3), Autosomal recessive
CARMIL2	99.99 %	610859	Immunodeficiency 58, 618131 (3), Autosomal recessive
CARS1	99.99 %	123859	Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive
CARS2	99.99 %	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
CARTPT	99.82 %	602606	{?Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive
CASK	98.95 %	300172	Intellectual developmental disorder, with or without nystagmus, 300422 (3), X-linked recessive; Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked; FG syndrome 4, 300422 (3), X-linked recessive
CASP10	99.85 %	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CASP12	99.98 %	608633	{Sepsis, susceptibility to} (3)
CASP14	99.86 %	605848	Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive
CASP2	99.99 %	600639	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653 (3), Autosomal recessive
CASP8	99.92 %	601763	{Breast cancer, protection against}, 114480 (3), Somatic mutation, Autosomal dominant; ?Caspase 8 lymphadenopathy syndrome, 607271 (3), Autosomal recessive; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Somatic mutation, Autosomal dominant
CASQ1	99.62 %	114250	Myopathy, vacuolar, with CASQ1 aggregates, 616231 (3), Autosomal dominant
CASQ2	94.39 %	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
CASR	99.99 %	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal dominant, Autosomal recessive; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CAST	99.94 %	114090	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive
CAT	99.89 %	115500	Acatalasemia, 614097 (3)
CATIP	99.99 %	619387	?Spermatogenic failure 54, 619379 (3), Autosomal recessive
CATSPER1	99.98 %	606389	Spermatogenic failure 7, 612997 (3), Autosomal recessive
CAV1	99.97 %	601047	Lipodystrophy, congenital generalized, type 3, 612526 (3), Autosomal recessive; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant; Lipodystrophy, familial partial, type 7, 606721 (3), Autosomal dominant
CAV3	100 %	601253	Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant
CAVIN1	100 %	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CBFB	99.79 %	121360	Cleidocranial dysplasia 2, 620099 (3), Autosomal dominant
CBL	99.95 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CBLB	99.55 %	604491	Autoimmune disease, multisystem, infantile-onset, 3, 620430 (3), Autosomal recessive
CBLIF	99.8 %	609342	Intrinsic factor deficiency, 261000 (3), Autosomal recessive
CBS	17.79 %	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
CBX2	100 %	602770	?46XY sex reversal 5, 613080 (3), Autosomal recessive
CC2D1A	99.98 %	610055	Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive
CC2D2A	99.95 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCBE1	99.52 %	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC103	99.68 %	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
CCDC115	99.9 %	613734	Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive
CCDC134	100 %	618788	Osteogenesis imperfecta, type XXII, 619795 (3), Autosomal recessive
CCDC146	99.13 %	619829	Spermatogenic failure 94, 620850 (3), Autosomal recessive
CCDC174	99.98 %	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
CCDC22	99.91 %	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
CCDC28B	99.99 %	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive
CCDC32	99.97 %	618941	Cardiofacioneurodevelopmental syndrome, 619123 (3), Autosomal recessive
CCDC34	99.75 %	612324	Spermatogenic failure 76, 620084 (3), Autosomal recessive
CCDC39	99.74 %	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
CCDC40	100 %	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
CCDC47	99.97 %	618260	Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive
CCDC50	99.88 %	611051	?Deafness, autosomal dominant 44, 607453 (3), Autosomal dominant
CCDC62	99.98 %	613481	?Spermatogenic failure 67, 619803 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CCDC65	99.8 %	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
CCDC78	100 %	614666	?Centronuclear myopathy 4, 614807 (3), Autosomal dominant
CCDC8	100 %	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
CCDC88A	99.48 %	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
CCDC88C	100 %	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
CCIN	99.99 %	603960	Spermatogenic failure 91, 620838 (3), Autosomal recessive
CCL11	99.96 %	601156	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {HIV1, resistance to}, 609423 (3)
CCL2	99.98 %	158105	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {HIV-1, resistance to}, 609423 (3); {Coronary artery disease, modifier of} (3); {Spina bifida, susceptibility to}, 182940 (3), Autosomal dominant
CCL3	100 %	182283	{HIV infection, resistance to}, 609423 (2)
CCL5	99.88 %	187011	{HIV-1 disease, rapid progression of}, 609423 (3); {HIV-1 disease, delayed progression of}, 609423 (3)
CCM2	99.93 %	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
CCN6	99.89 %	603400	Progressive pseudorheumatoid dysplasia, 208230 (3), Autosomal recessive
CCND1	100 %	168461	{von Hippel-Lindau syndrome, modifier of}, 193300 (3), Autosomal dominant; {Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant; {Multiple myeloma, susceptibility to}, 254500 (3), Somatic mutation
CCND2	100 %	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
CCNF	99.99 %	600227	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141 (3), Autosomal dominant
CCNK	99.93 %	603544	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant
CCNO	100 %	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
CCNQ	99.98 %	300708	STAR syndrome, 300707 (3), X-linked dominant
CCR2	100 %	601267	{HIV infection, susceptibility/resistance to}, 609423 (3); Polycystic lung disease, 219600 (3), Autosomal recessive
CCR5	99.97 %	601373	{HIV infection, susceptibility/resistance to}, 609423 (3); {Diabetes mellitus, insulin-dependent, 22}, 612522 (3); {Hepatitis C virus, resistance to}, 609532 (3); {West nile virus, susceptibility to}, 610379 (3)
CCT5	99.99 %	610150	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
CD151	100 %	602243	[Blood group, Raph], 179620 (3); Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057 (3), Autosomal recessive
CD164	99.97 %	603356	?Deafness, autosomal dominant 66, 616969 (3), Autosomal dominant
CD19	99.98 %	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD207	99.92 %	604862	[?Birbeck granule deficiency], 613393 (3)
CD209	100 %	604672	{HIV type 1, susceptibility to}, 609423 (3); {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {Dengue fever, protection against}, 614371 (3)
CD244	99.64 %	605554	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
CD247	99.79 %	186780	?Immunodeficiency 25, 610163 (3), Autosomal recessive
CD27	99.95 %	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
CD28	99.99 %	186760	?Immunodeficiency 123 with HPV-related verrucosis, 620901 (3), Autosomal recessive
CD2AP	99.69 %	604241	Glomerulosclerosis, focal segmental, 3, 607832 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CD320	100 %	606475	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (3), Autosomal recessive
CD36	99.62 %	173510	Platelet glycoprotein IV deficiency, 608404 (3), Autosomal recessive; {Coronary heart disease, susceptibility to, 7}, 610938 (3); {Malaria, cerebral, susceptibility to}, 611162 (3); {Malaria, cerebral, reduced risk of}, 611162 (3)
CD3D	100 %	186790	Immunodeficiency 19, severe combined, 615617 (3), Autosomal recessive
CD3E	100 %	186830	Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive
CD3G	100 %	186740	Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive
CD4	100 %	186940	Immunodeficiency 79, 619238 (3), Autosomal recessive; OKT4 epitope deficiency, 613949 (3)
CD40	100 %	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
CD40LG	99.88 %	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
CD44	99.95 %	107269	[Blood group, Indian system], 609027 (3)
CD46	99.86 %	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal dominant, Autosomal recessive
CD55	74.12 %	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive
CD59	100 %	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
CD70	99.99 %	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive
CD79A	99.97 %	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
CD79B	99.93 %	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
CD81	99.97 %	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
CD8A	99.97 %	186910	Immunodeficiency 116, 608957 (3), Autosomal recessive
CD96	99.91 %	606037	C syndrome, 211750 (3), Autosomal dominant
CDAN1	100 %	607465	Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive
CDC14A	99.04 %	603504	Deafness, autosomal recessive 32, with or without immotile sperm, 608653 (3), Autosomal recessive
CDC20	99.89 %	603618	Oocyte/zygote/embryo maturation arrest 14, 620276 (3), Autosomal recessive
CDC40	99.64 %	605585	?Pontocerebellar hypoplasia, type 15, 619302 (3), Autosomal recessive
CDC42	98.05 %	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
CDC42BPB	99.99 %	614062	Chilton-Okur-Chung neurodevelopmental syndrome, 619841 (3), Autosomal dominant
CDC45	99.77 %	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
CDC6	99.85 %	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
CDC73	99.6 %	607393	Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant; Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3); Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant
CDCA7	99.88 %	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
CDH1	99.98 %	192090	Ovarian cancer, somatic, 167000 (3); Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); Breast cancer, lobular, somatic, 114480 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CDH11	99.94 %	600023	Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elsahy-Waters syndrome, 211380 (3), Autosomal recessive
CDH15	99.98 %	114019	Intellectual developmental disorder, autosomal dominant 3, 612580 (3), Autosomal dominant
CDH2	99.84 %	114020	Arrhythmogenic right ventricular dysplasia 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant
CDH23	99.95 %	605516	Usher syndrome, type 1D, 601067 (3), Digenic recessive, Autosomal recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D/F digenic, 601067 (3), Digenic recessive, Autosomal recessive; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive
CDH3	99.98 %	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
CDHR1	99.72 %	609502	Macular dystrophy, retinal, 613660 (3), Autosomal recessive; Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
CDIN1	98.04 %	615626	Dyserythropoietic anemia, congenital, type Ib, 615631 (3), Autosomal recessive
CDK10	99.98 %	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
CDK13	99.83 %	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
CDK19	99.26 %	614720	Developmental and epileptic encephalopathy 87, 618916 (3), Autosomal dominant
CDK4	100 %	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
CDK5	99.95 %	123831	?Lissencephaly 7 with cerebellar hypoplasia, 616342 (3), Autosomal recessive
CDK5RAP2	99.97 %	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDK6	99.47 %	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
CDK8	99.81 %	603184	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant
CDKL5	99.88 %	300203	Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant
CDKN1B	100 %	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
CDKN1C	100 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDKN2A	100 %	600160	{Melanoma and neural system tumor syndrome}, 155755 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 2}, 155601 (3), Autosomal dominant; {Melanoma-pancreatic cancer syndrome}, 606719 (3), Autosomal dominant
CDON	99.99 %	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant
CDSN	99.94 %	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
CDT1	100 %	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
CEACAM16	100 %	614591	Deafness, autosomal dominant 4B, 614614 (3), Autosomal dominant; Deafness, autosomal recessive 113, 618410 (3), Autosomal recessive
CEBPA	100 %	116897	Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant
CEBPE	100 %	600749	?Immunodeficiency 108 with autoinflammation, 260570 (3), Autosomal recessive; Specific granule deficiency, 245480 (3), Autosomal dominant, Autosomal recessive
CEL	97.13 %	114840	Maturity-onset diabetes of the young, type VIII, 609812 (3), Autosomal dominant
CELA2A	100 %	609443	Abdominal obesity-metabolic syndrome 4, 618620 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CELF2	99.99 %	602538	Developmental and epileptic encephalopathy 97, 619561 (3), Autosomal dominant
CELSR1	99.99 %	604523	Lymphatic malformation 9, 619319 (3), Autosomal dominant
CENATAC	99.97 %	620142	?Mosaic variegated aneuploidy syndrome 4, 620153 (3), Autosomal recessive
CENPE	99.78 %	117143	?Microcephaly 13, primary, autosomal recessive, 616051 (3), Autosomal recessive
CENPF	99.97 %	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CENPJ	99.92 %	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
CENPT	100 %	611510	?Short stature and microcephaly with genital anomalies, 618702 (3), Autosomal recessive
CEP104	99.99 %	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
CEP112	99.88 %	618980	Spermatogenic failure 44, 619044 (3), Autosomal recessive
CEP120	99.9 %	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
CEP135	99.82 %	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
CEP152	99.93 %	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
CEP164	99.99 %	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
CEP19	99.99 %	615586	Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive
CEP250	99.93 %	609689	Cone-rod dystrophy and hearing loss 2, 618358 (3), Autosomal recessive
CEP290	98.1 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CEP295	99.94 %	617728	Seckel syndrome 11, 620767 (3), Autosomal recessive
CEP41	99.99 %	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP43	99.8 %	605392	Myeloproliferative disorder, 605392 (2)
CEP55	99.92 %	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
CEP57	99.92 %	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
CEP63	94.73 %	614724	?Seckel syndrome 6, 614728 (3), Autosomal recessive
CEP78	99.95 %	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
CEP83	98.68 %	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CEP85L	100 %	618865	Lissencephaly 10, 618873 (3), Autosomal dominant
CERKL	99.91 %	608381	Retinitis pigmentosa 26, 608380 (3), Autosomal recessive
CERS1	100 %	606919	Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive
CERS3	99.85 %	615276	Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive
CERT1	99.66 %	604677	Intellectual developmental disorder, autosomal dominant 34, 616351 (3), Autosomal dominant
CES1	80.16 %	114835	Drug metabolism, altered, CES1-related, 618057 (3), Autosomal dominant
CETP	100 %	118470	[High density lipoprotein cholesterol level QTL 10], 143470 (3), Autosomal dominant; Hyperalphalipoproteinemia, 143470 (3), Autosomal dominant
CFAP251	99.98 %	618146	Spermatogenic failure 33, 618152 (3), Autosomal recessive
CFAP298	99.96 %	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
CFAP300	99.47 %	618058	Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CFAP410	100 %	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
CFAP418	100 %	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive
CFAP43	99.88 %	617558	Hydrocephalus, normal pressure, 1, 236690 (3), Autosomal dominant; Spermatogenic failure 19, 617592 (3), Autosomal recessive
CFAP44	99.75 %	617559	Spermatogenic failure 20, 617593 (3), Autosomal recessive
CFAP45	99.97 %	605152	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608 (3), Autosomal recessive
CFAP47	99.21 %	301057	Spermatogenic failure, X-linked 3, 301059 (3), X-linked recessive
CFAP52	99.92 %	609804	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607 (3), Autosomal recessive
CFAP53	99.95 %	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
CFAP57	98.9 %	614259	Spermatogenic failure 95, 620917 (3), Autosomal recessive
CFAP58	99.96 %	619129	Spermatogenic failure 49, 619144 (3), Autosomal recessive
CFAP61	99.98 %	620381	Spermatogenic failure 84, 620409 (3), Autosomal recessive
CFAP65	99.99 %	614270	Spermatogenic failure 40, 618664 (3), Autosomal recessive
CFAP69	99.09 %	617949	Spermatogenic failure 24, 617959 (3), Autosomal recessive
CFAP70	99.74 %	618661	?Spermatogenic failure 41, 618670 (3), Autosomal recessive
CFAP74	99.98 %	620187	Ciliary dyskinesia, primary, 49, without situs inversus, 620197 (3), Autosomal recessive
CFAP91	99.97 %	609910	Spermatogenic failure 51, 619177 (3), Autosomal recessive
CFB	99.97 %	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
CFC1	21.93 %	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
CFD	99.99 %	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
CFH	99.12 %	134370	{Macular degeneration, age-related, 4}, 610698 (3), Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal dominant, Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal dominant, Autosomal recessive
CFHR1	84.44 %	134371	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal dominant, Autosomal recessive
CFHR3	91.62 %	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal dominant, Autosomal recessive
CFHR5	99.68 %	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
CFI	99.87 %	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive
CFL2	99.67 %	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
CFP	99.96 %	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CFTR	99.45 %	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
CHAMP1	99.99 %	616327	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579 (3), Autosomal dominant
CHAT	99.74 %	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive
CHCHD10	100 %	615903	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant; Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant
CHCHD2	99.89 %	616244	Parkinson disease 22, autosomal dominant, 616710 (3), Autosomal dominant
CHD1	99.46 %	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
CHD2	99.97 %	602119	Developmental and epileptic encephalopathy 94, 615369 (3), Autosomal dominant
CHD3	99.06 %	602120	Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant
CHD4	99.99 %	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD5	99.97 %	610771	Parenti-Mignot neurodevelopmental syndrome, 619873 (3), Autosomal dominant
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHD8	99.96 %	610528	Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant
CHEK1	99.89 %	603078	Oocyte/zygote/embryo maturation arrest 21, 620610 (3), Autosomal dominant
CHEK2	92.84 %	604373	Prostate cancer, somatic, 176807 (3); Osteosarcoma, somatic, 259500 (3); Tumor predisposition syndrome 4, breast/prostate/colorectal, 609265 (3)
CHI3L1	100 %	601525	{Asthma-related traits, susceptibility to, 7}, 611960 (3); {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
CHIC2	99.72 %	604332	{Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
CHIT1	99.94 %	600031	[Chitotriosidase deficiency], 614122 (3), Autosomal recessive
CHKA	99.95 %	118491	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023 (3), Autosomal recessive
CHKB	100 %	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHM	96.46 %	300390	Choroideremia, 303100 (3), X-linked
CHMP1A	100 %	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
CHMP2B	99.8 %	609512	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795 (3), Autosomal dominant
CHMP4B	99.96 %	610897	Cataract 31, multiple types, 605387 (3), Autosomal dominant
CHN1	99.83 %	118423	Duane retraction syndrome 2, 604356 (3), Autosomal dominant
CHP1	99.9 %	606988	?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive
CHRDL1	99.94 %	300350	Megalocornea 1, X-linked, 309300 (3), X-linked recessive
CHRM3	100 %	118494	Prune belly syndrome, 100100 (3), Autosomal recessive
CHRNA1	99.82 %	100690	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal dominant, Autosomal recessive; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
CHRNA2	99.98 %	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CHRNA3	99.95 %	118503	{Lung cancer susceptibility 2}, 612052 (3); Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 (3), Autosomal recessive
CHRNA4	100 %	118504	{Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant
CHRNA5	99.92 %	118505	{Nicotine dependence, susceptibility to}, 612052 (3); {Lung cancer susceptibility 2}, 612052 (3)
CHRNB1	99.99 %	100710	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant
CHRNB2	99.99 %	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CHRND	100 %	100720	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant
CHRNE	100 %	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal dominant, Autosomal recessive; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive
CHRNG	100 %	100730	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Escobar syndrome, 265000 (3), Autosomal recessive
CHST11	99.99 %	610128	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167 (3), Autosomal recessive
CHST14	100 %	608429	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive
CHST3	100 %	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive
CHST6	100 %	605294	Macular corneal dystrophy, 217800 (3), Autosomal recessive
CHSY1	99.99 %	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
CHUK	99.83 %	600664	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 (3), Autosomal recessive; ?Cocoon syndrome, 613630 (3), Autosomal recessive
CIB1	99.92 %	602293	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267 (3), Autosomal recessive
CIB2	100 %	605564	Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type IJ, 614869 (3), Autosomal recessive
CIBAR1	99.94 %	617273	?Polydactyly, postaxial, type A9, 618219 (3), Autosomal recessive
CIC	98.32 %	612082	Intellectual developmental disorder, autosomal dominant 45, 617600 (3), Autosomal dominant
CIDEC	100 %	612120	?Lipodystrophy, familial partial, type 5, 615238 (3), Autosomal recessive
CIITA	99.99 %	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); MHC class II deficiency 1, 209920 (3), Autosomal recessive
CILK1	99.69 %	612325	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
CILP	100 %	603489	{Lumbar disc disease, susceptibility to}, 603932 (3)
CISD2	96.27 %	611507	Wolfram syndrome 2, 604928 (3), Autosomal recessive
CISH	100 %	602441	{Malaria, susceptibility to}, 611162 (3); {Bacteremia, susceptibility to}, 614383 (3); {Tuberculosis, susceptibility to}, 607948 (3)
CIT	99.99 %	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CITED2	100 %	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
CKAP2L	99.54 %	616174	Filippi syndrome, 272440 (3), Autosomal recessive
CLCC1	93.17 %	617539	Retinitis pigmentosa 32, 609913 (3), Autosomal recessive
CLCF1	100 %	607672	Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive
CLCN1	100 %	118425	Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia levior, 160800 (3), Autosomal dominant
CLCN2	100 %	600570	Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant
CLCN3	99.95 %	600580	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 (3), Autosomal recessive; Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 (3), Autosomal dominant
CLCN4	99.98 %	302910	Raynaud-Claes syndrome, 300114 (3), X-linked dominant
CLCN5	99.67 %	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Dent disease 1, 300009 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
CLCN6	100 %	602726	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173 (3), Autosomal dominant
CLCN7	99.99 %	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
CLCNKA	99.98 %	602024	Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLCNKB	99.98 %	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN1	99.99 %	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
CLDN10	99.97 %	617579	HELIX syndrome, 617671 (3), Autosomal recessive
CLDN11	100 %	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
CLDN14	100 %	605608	Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive
CLDN16	99.98 %	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	99.02 %	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLDN2	100 %	300520	?Azoospermia, obstructive, with nephrolithiasis, 301060 (3), X-linked recessive
CLDN9	100 %	615799	Deafness, autosomal recessive 116, 619093 (3), Autosomal recessive
CLEC1A	99.97 %	606782	{Aspergillosis, susceptibility to}, 614079 (3)
CLEC3B	99.94 %	187520	Macular dystrophy, retinal, 4, 619977 (3), Autosomal dominant
CLEC7A	99.98 %	606264	Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3)
CLIC5	99.97 %	607293	?Deafness, autosomal recessive 103, 616042 (3), Autosomal recessive
CLMP	100 %	611693	Congenital short bowel syndrome, 615237 (3), Autosomal recessive
CLN3	99.92 %	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CLN6	100 %	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
CLN8	100 %	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive
CLP1	99.98 %	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
CLPB	99.97 %	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
CLPP	99.99 %	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
CLPX	99.88 %	615611	?Protoporphyria, erythropoietic, 2, 618015 (3), Autosomal dominant
CLRN1	99.99 %	606397	Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3)
CLRN2	100 %	618988	Deafness, autosomal recessive 117, 619174 (3), Autosomal recessive
CLTC	99.56 %	118955	Intellectual developmental disorder, autosomal dominant 56, 617854 (3), Autosomal dominant
CNBP	100 %	116955	Myotonic dystrophy 2, 602668 (3), Autosomal dominant
CNGA1	99.6 %	123825	Retinitis pigmentosa 49, 613756 (3), Autosomal recessive
CNGA3	99.95 %	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
CNGB1	97.53 %	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
CNGB3	99.96 %	605080	Achromatopsia 3, 262300 (3), Autosomal recessive
CNKSR2	99.54 %	300724	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked
CNNM2	99.94 %	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 (3), Autosomal dominant, Autosomal recessive
CNNM4	99.93 %	607805	Jalili syndrome, 217080 (3), Autosomal recessive
CNOT1	99.83 %	604917	Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant
CNOT2	99 %	604909	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant
CNOT3	99.99 %	604910	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant
CNP	99.98 %	123830	?Leukodystrophy, hypomyelinating, 20, 619071 (3), Autosomal recessive
CNPY3	99.98 %	610774	Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive
CNTN1	99.35 %	600016	Congenital myopathy 12, 612540 (3), Autosomal recessive
CNTN2	99.95 %	190197	Epilepsy, early-onset, 5, with or without developmental delay, 615400 (3), Autosomal recessive
CNTNAP1	99.98 %	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
CNTNAP2	99.99 %	604569	Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3)
COA3	99.99 %	614775	?Mitochondrial complex IV deficiency, nuclear type 14, 619058 (3), Autosomal recessive
COA5	98.65 %	613920	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COA6	99.98 %	614772	Mitochondrial complex IV deficiency, nuclear type 13, 616501 (3), Autosomal recessive
COA7	99.91 %	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
COA8	99.94 %	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
COASY	99.98 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COCH	100 %	603196	Deafness, autosomal dominant 9, 601369 (3), Autosomal dominant; ?Deafness, autosomal recessive 110, 618094 (3), Autosomal recessive
COG1	100 %	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
COG2	99.73 %	606974	?Congenital disorder of glycosylation, type IIq, 617395 (3), Autosomal recessive
COG3	99.93 %	606975	Congenital disorder of glycosylation, type IIbb, 620546 (3), Autosomal recessive
COG4	99.96 %	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
COG5	99.92 %	606821	Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive
COG6	99.86 %	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
COG7	99.74 %	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
COG8	100 %	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
COL10A1	100 %	120110	Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant
COL11A1	90.72 %	120280	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3)
COL11A2	99.99 %	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal dominant, Autosomal recessive; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant
COL12A1	99.85 %	120320	Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3), Autosomal recessive
COL13A1	99.95 %	120350	Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive
COL17A1	99.98 %	113811	Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional 4, intermediate, 619787 (3), Autosomal recessive
COL18A1	99.99 %	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
COL1A1	99.9 %	120150	Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COL1A2	99.34 %	120160	Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant
COL25A1	99.87 %	610004	Fibrosis of extraocular muscles, congenital, 5, 616219 (3), Autosomal recessive
COL27A1	99.97 %	608461	Steel syndrome, 615155 (3), Autosomal recessive
COL2A1	99.87 %	120140	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant
COL3A1	99.87 %	120180	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive
COL4A1	99.99 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COL4A2	99.98 %	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
COL4A3	99.94 %	120070	Alport syndrome 3A, autosomal dominant, 104200 (3), Autosomal dominant; Hematuria, benign familial, 2, 620320 (3), Autosomal dominant; Alport syndrome 3B, autosomal recessive, 620536 (3)
COL4A4	99.95 %	120131	Hematuria, familial benign, 1, 141200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive
COL4A5	99.64 %	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant
COL4A6	99.97 %	303631	?Deafness, X-linked 6, 300914 (3), X-linked recessive
COL5A1	99.99 %	120215	Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant; Fibromuscular dysplasia, multifocal, 619329 (3), Autosomal dominant
COL5A2	99.86 %	120190	Ehlers-Danlos syndrome, classic type, 2, 130010 (3), Autosomal dominant
COL6A1	99.99 %	120220	Ullrich congenital muscular dystrophy 1A, 254090 (3), Autosomal dominant, Autosomal recessive; Bethlem myopathy 1A, 158810 (3), Autosomal dominant
COL6A2	100 %	120240	?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1B, 620727 (3), Autosomal dominant, Autosomal recessive; Bethlem myopathy 1B, 620725 (3), Autosomal dominant, Autosomal recessive
COL6A3	99.99 %	120250	Bethlem myopathy 1C, 620726 (3), Autosomal dominant, Autosomal recessive; Ullrich congenital muscular dystrophy 1C, 620728 (3), Autosomal dominant, Autosomal recessive; Dystonia 27, 616411 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COL7A1	99.99 %	120120	Nail disorder, nonsyndromic congenital, 8, 607523 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, autosomal recessive, 226600 (3), Autosomal recessive; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal dominant, Autosomal recessive; Epidermolysis bullosa dystrophica, autosomal dominant, 131750 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal dominant, Autosomal recessive; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal dominant, Autosomal recessive; Epidermolysis bullosa dystrophica, localisata variant, 226600 (3), Autosomal recessive
COL8A2	99.94 %	120252	Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant
COL9A1	99.91 %	120210	Stickler syndrome, type IV, 614134 (3), Autosomal recessive; ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant
COL9A2	98.76 %	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
COL9A3	99.99 %	120270	{Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; Stickler syndrome, type VI, 620022 (3), Autosomal recessive
COLEC10	99.97 %	607620	3MC syndrome 3, 248340 (3), Autosomal recessive
COLEC11	100 %	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
COLGALT1	99.82 %	617531	Brain small vessel disease 3, 618360 (3), Autosomal recessive
COLQ	99.98 %	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
COMP	100 %	600310	Pseudoachondroplasia, 177170 (3), Autosomal dominant; Carpal tunnel syndrome 2, 619161 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant
COMT	99.96 %	116790	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Panic disorder, susceptibility to}, 167870 (3), ?Autosomal dominant
COPA	99.61 %	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
COPB1	99.87 %	600959	Baralle-Macken syndrome, 619255 (3), Autosomal recessive
COPB2	99.83 %	606990	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive
COQ2	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ4	100 %	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive; Spastic ataxia 10, autosomal recessive, 620666 (3), Autosomal recessive
COQ5	99.95 %	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
COQ6	99.94 %	614647	Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive
COQ7	100 %	601683	Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 (3), Autosomal recessive
COQ8A	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ8B	99.94 %	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
COQ9	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
CORIN	99.96 %	605236	?Cardiomyopathy, familial hypertrophic, 30, atrial, 620734 (3), Autosomal recessive; Preeclampsia/eclampsia 5, 614595 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CORO1A	91.71 %	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
COX10	99.99 %	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
COX11	99.72 %	603648	Mitochondrial complex IV deficiency, nuclear type 23, 620275 (3), Autosomal recessive
COX14	99.99 %	614478	?Mitochondrial complex IV deficiency, nuclear type 10, 619053 (3), Autosomal recessive
COX15	100 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
COX16	100 %	618064	Mitochondrial complex IV deficiency, nuclear type 22, 619355 (3), Autosomal recessive
COX20	99.67 %	614698	Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive
COX4I1	100 %	123864	Mitochondrial complex IV deficiency, nuclear type 16, 619060 (3), Autosomal recessive
COX4I2	100 %	607976	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 (3), Autosomal recessive
COX5A	99.7 %	603773	Mitochondrial complex IV deficiency, nuclear type 20, 619064 (3), Autosomal recessive
COX6A1	99.98 %	602072	Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive
COX6A2	99.75 %	602009	Mitochondrial complex IV deficiency, nuclear type 18, 619062 (3), Autosomal recessive
COX6B1	100 %	124089	Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive
COX7B	99.86 %	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
COX8A	99.73 %	123870	?Mitochondrial complex IV deficiency, nuclear type 15, 619059 (3), Autosomal recessive
CP	99.95 %	117700	Aceruloplasminemia, 604290 (3), Autosomal recessive
CPA6	99.99 %	609562	Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal dominant, Autosomal recessive
CPAMD8	99.97 %	608841	Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive
CPE	99.93 %	114855	BDV syndrome, 619326 (3), Autosomal recessive
CPLANE1	99.81 %	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
CPLX1	100 %	605032	Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive
CPN1	99.96 %	603103	Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive
CPOX	99.99 %	612732	Coproporphyria, 121300 (3), Autosomal dominant, Autosomal recessive; Harderoporphyria, 618892 (3), Autosomal recessive
CPS1	99.91 %	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
CPSF1	99.99 %	606027	Myopia 27, 618827 (3), Autosomal dominant
CPSF3	99.93 %	606029	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 (3), Autosomal recessive
CPT1A	99.98 %	600528	CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive
CPT1C	99.99 %	608846	?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CPT2	99.65 %	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal dominant, Autosomal recessive; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal dominant, Autosomal recessive
CR1	66.67 %	120620	[Blood group, Knops system], 607486 (3); {Malaria, severe, resistance to}, 611162 (3)
CR2	99.97 %	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); ?Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
CRADD	99.9 %	603454	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
CRAT	99.99 %	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
CRB1	99.73 %	604210	Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant
CRB2	99.95 %	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
CRBN	99.97 %	609262	Intellectual developmental disorder, autosomal recessive 2, 607417 (3), Autosomal recessive
CREB1	99.95 %	123810	Histiocytoma, angiomatoid fibrous, somatic, 612160 (3)
CREB3L1	99.88 %	616215	Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive
CREB3L3	99.98 %	611998	Hypertriglyceridemia 2, 619324 (3), Autosomal dominant
CREBBP	99.97 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRELD1	99.99 %	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; Jeffries-Lakhani neurodevelopmental syndrome, 620771 (3), Autosomal recessive; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
CRIPT	99.96 %	604594	Rothmund-Thomson syndrome, type 3, 615789 (3), Autosomal recessive
CRLF1	99.99 %	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CRLS1	99.93 %	608188	Combined oxidative phosphorylation deficiency 57, 620167 (3), Autosomal recessive
CRPPA	99.98 %	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
CRTAP	99.98 %	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
CRTC1	99.96 %	607536	Mucoepidermoid salivary gland carcinoma (3)
CRX	99.96 %	602225	Leber congenital amaurosis 7, 613829 (3); Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant
CRY1	99.86 %	601933	{Delayed sleep phase disorder, susceptibility to}, 614163 (3), Autosomal dominant
CRYAA	19.49 %	123580	Cataract 9, multiple types, 604219 (3), Autosomal dominant, Autosomal recessive
CRYAB	100 %	123590	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant
CRYBA1	99.99 %	123610	Cataract 10, multiple types, 600881 (3), Autosomal dominant
CRYBA2	100 %	600836	?Cataract 42, 115900 (3), Autosomal dominant
CRYBA4	100 %	123631	Cataract 23, 610425 (3), Autosomal dominant
CRYBB1	99.46 %	600929	Cataract 17, multiple types, 611544 (3), Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CRYBB2	99.94 %	123620	Cataract 3, multiple types, 601547 (3), Autosomal dominant
CRYBB3	99.99 %	123630	Cataract 22, 609741 (3), Autosomal dominant, Autosomal recessive
CRYGB	99.99 %	123670	Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant
CRYGC	100 %	123680	Cataract 2, multiple types, 604307 (3), Autosomal dominant
CRYGD	100 %	123690	Cataract 4, multiple types, 115700 (3), Autosomal dominant
CRYGS	100 %	123730	Cataract 20, multiple types, 116100 (3), Autosomal dominant
CRYM	99.94 %	123740	Deafness, autosomal dominant 40, 616357 (3), Autosomal dominant
CSF1R	99.92 %	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant
CSF2RA	93.86 %	425000	*No OMIM phenotypes*
CSF2RB	100 %	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
CSF3R	99.97 %	138971	Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive; ?Neutrophilia, hereditary, 162830 (3), Autosomal dominant
CSGALNACT1	100 %	616615	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive
CSH1	78.25 %	150200	[Placental lactogen deficiency] (1)
CSNK1D	99.99 %	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
CSNK2A1	99.96 %	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
CSNK2B	99.65 %	115441	Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant
CSPP1	98.31 %	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CSRP3	100 %	600824	?Cardiomyopathy, dilated, 1M, 607482 (3); Cardiomyopathy, hypertrophic, 12, 612124 (3), Autosomal dominant
CST3	100 %	604312	{Macular degeneration, age-related, 11}, 611953 (3); Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant
CST6	99.99 %	601891	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535 (3), Autosomal recessive
CSTA	99.83 %	184600	Peeling skin syndrome 4, 607936 (3), Autosomal recessive
CSTB	100 %	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CSTF2	99.81 %	300907	?Intellectual developmental disorder, X-linked 113, 301116 (3), X-linked recessive
CT55	54.67 %	301105	?Spermatogenic failure, X-linked, 7, 301106 (3), X-linked recessive
CTBP1	99.98 %	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
CTC1	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTCF	99.87 %	604167	Intellectual developmental disorder, autosomal dominant 21, 615502 (3), Autosomal dominant
CTDP1	99.97 %	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CTH	93.58 %	607657	Cystathioninuria, 219500 (3), Autosomal recessive
CTHRC1	99.9 %	610635	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
CTLA4	99.99 %	123890	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Celiac disease, susceptibility to, 3}, 609755 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
CTNNA1	99.98 %	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CTNNA2	99.84 %	114025	Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive
CTNNA3	99.96 %	607667	Arrhythmogenic right ventricular dysplasia 13, 615616 (3), Autosomal dominant
CTNNB1	99.95 %	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3)
CTNNBL1	100 %	611537	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846 (3), Autosomal recessive
CTNND1	99.92 %	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
CTNS	100 %	606272	Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive
CTPS1	98.63 %	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
CTRC	100 %	601405	{Pancreatitis, chronic, susceptibility to}, 167800 (3), Autosomal dominant
CTSA	99.98 %	613111	Galactosialidosis, 256540 (3), Autosomal recessive
CTSB	99.99 %	116810	Keratolytic winter erythema, 148370 (4), Autosomal dominant
CTSC	99.97 %	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
CTSD	100 %	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CTSF	99.96 %	603539	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive
CTSK	99.31 %	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
CTU2	99.91 %	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive
CUBN	99.99 %	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
CUL3	99.76 %	603136	Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
CUL4B	99.67 %	300304	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive
CUL7	100 %	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
CUX1	99.37 %	116896	Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant
CUX2	99.96 %	610648	Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant
CWC27	99.67 %	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
CWF19L1	99.91 %	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
CX3CR1	100 %	601470	{Rapid progression to AIDS from HIV1 infection}, 609423 (3); {Macular degeneration, age-related, 12}, 613784 (3); {Coronary artery disease, resistance to}, 607339 (3)
CXCL12	99.97 %	600835	{AIDS, resistance to}, 609423 (3)
CXCR1	100 %	146929	{AIDS, slow progression to}, 609423 (3)
CXCR2	100 %	146928	?WHIM syndrome 2, 619407 (3), Autosomal recessive
CXCR4	99.98 %	162643	WHIM syndrome 1, 193670 (3), Autosomal dominant; Myelokathexis, isolated, 193670 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CYB561	99.99 %	600019	Orthostatic hypotension 2, 618182 (3), Autosomal recessive
CYB5A	99.98 %	613218	Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive
CYB5R3	99.93 %	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
CYBA	99.96 %	608508	Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive
CYBB	99.87 %	300481	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive
CYBC1	100 %	618334	Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive
CYC1	99.86 %	123980	Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive
CYCS	88 %	123970	Thrombocytopenia 4, 612004 (3), Autosomal dominant
CYFIP2	100 %	606323	Developmental and epileptic encephalopathy 65, 618008 (3), Autosomal dominant
CYLC1	99.71 %	300768	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119 (3), X-linked
CYLD	99.46 %	605018	Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Cylindromatosis, familial, 132700 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant; ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132 (3), Autosomal dominant
CYP11A1	99.99 %	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	100 %	610613	Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive
CYP11B2	100 %	124080	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Aldosterone to renin ratio raised (3); {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive
CYP17A1	100 %	609300	17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive
CYP19A1	99.99 %	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
CYP1B1	100 %	601771	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive; Anterior segment dysgenesis 6, multiple subtypes, 617315 (3), Autosomal recessive
CYP21A2	99.85 %	613815	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
CYP24A1	100 %	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
CYP26B1	100 %	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3)
CYP26C1	99.92 %	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP27B1	99.98 %	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
CYP2A6	99.98 %	122720	{Lung cancer, resistance to}, 211980 (3), Somatic mutation, Autosomal dominant; Coumarin resistance, 122700 (3), Autosomal dominant; {Nicotine addiction, protection from}, 188890 (3)
CYP2B6	99.99 %	123930	{Efavirenz central nervous system toxicity, susceptibility to}, 614546 (3); Efavirenz, poor metabolism of, 614546 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CYP2C19	100 %	124020	Proguanil poor metabolizer, 609535 (3), Autosomal recessive; Mephenytoin poor metabolizer, 609535 (3), Autosomal recessive; Clopidogrel, impaired responsiveness to, 609535 (3), Autosomal recessive; Omeprazole poor metabolizer, 609535 (3), Autosomal recessive
CYP2C8	99.79 %	601129	{Drug metabolism, altered, CYP2C8-related}, 618018 (3)
CYP2C9	99.95 %	601130	Warfarin sensitivity, 122700 (3), Autosomal dominant; Tolbutamide poor metabolizer (3)
CYP2D6	99.95 %	124030	{Codeine sensitivity}, 608902 (3), Autosomal recessive; {Debrisoquine sensitivity}, 608902 (3), Autosomal recessive
CYP2R1	99.96 %	608713	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081 (3), Autosomal recessive
CYP2U1	99.99 %	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
CYP3A4	99.73 %	124010	Vitamin D-dependent rickets, type 3, 619073 (3), Autosomal dominant
CYP3A5	99.71 %	605325	{Hypertension, salt-sensitive essential, susceptibility to}, 145500 (3), Multifactorial
CYP4F22	99.97 %	611495	Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive
CYP4V2	99.98 %	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
CYP7B1	99.82 %	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
D2HGDH	100 %	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
DAAM2	99.99 %	606627	Nephrotic syndrome, type 24, 619263 (3), Autosomal recessive
DAB1	97.87 %	603448	Spinocerebellar ataxia 37, 615945 (3), Autosomal dominant
DACT1	99.99 %	607861	Townes-Brocks syndrome 2, 617466 (3), Autosomal dominant
DAG1	100 %	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive
DAGLA	99.93 %	614015	Neuroocular syndrome 2, paroxysmal type, 168885 (3), Autosomal dominant
DALRD3	99.99 %	618904	?Developmental and epileptic encephalopathy 86, 618910 (3), Autosomal recessive
DAOA	100 %	607408	{Schizophrenia}, 181500 (2), Autosomal dominant
DARS1	98.85 %	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
DARS2	98.31 %	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DAW1	99.9 %	620279	Ciliary dyskinesia, primary, 52, 620570 (3), Autosomal recessive
DAZL	99.49 %	601486	{Spermatogenic failure, susceptibility to} (3)
DBH	99.99 %	609312	Orthostatic hypotension 1, due to DBH deficiency, 223360 (3), Autosomal recessive
DBR1	99.92 %	607024	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510 (3), Autosomal recessive; {Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441 (3), Autosomal recessive
DBT	94.51 %	248610	Maple syrup urine disease, type II, 620699 (3), Autosomal recessive
DCAF17	99.84 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCAF8	99.83 %	615820	?Giant axonal neuropathy 2, autosomal dominant, 610100 (3), Autosomal dominant
DCC	99.96 %	120470	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DCDC2	99.96 %	605755	Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
DCHS1	100 %	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
DCLRE1B	99.91 %	609683	Dyskeratosis congenita, autosomal recessive 8, 620133 (3), Autosomal recessive
DCLRE1C	99.79 %	605988	Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
DCN	99.42 %	125255	Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant
DCPS	99.98 %	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DCT	99.98 %	191275	Oculocutaneous albinism, type VIII, 619165 (3), Autosomal recessive
DCTN1	99.98 %	601143	Perry syndrome, 168605 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal dominant, Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641 (3), Autosomal dominant
DCX	99.99 %	300121	Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked
DCXR	100 %	608347	[Pentosuria], 260800 (3), Autosomal recessive
DDB1	99.96 %	600045	White-Kernohan syndrome, 619426 (3), Autosomal dominant
DDB2	100 %	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
DDC	99.67 %	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DDHD1	99.93 %	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
DDHD2	99.97 %	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDOST	99.93 %	602202	Congenital disorder of glycosylation, type Ir, 614507 (3), Autosomal recessive
DDR2	99.58 %	191311	Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant; Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive
DDRGK1	100 %	616177	Spondyloepimetaphyseal dysplasia, Shohat type, 602557 (3), Autosomal recessive
DDX11	99.74 %	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
DDX3X	99.01 %	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
DDX41	99.99 %	608170	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871 (3), Autosomal dominant
DDX58	99.84 %	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DDX59	99.67 %	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DDX6	99.88 %	600326	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant
DEAF1	99.9 %	602635	Vulto-van Silfout-de Vries syndrome, 615828 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 (3), Autosomal recessive
DEF6	100 %	610094	Immunodeficiency 87 and autoimmunity, 619573 (3), Autosomal recessive
DEGS1	99.99 %	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DEK	99.14 %	125264	Leukemia, acute nonlymphocytic, 125264 (2)
DENND5A	99.99 %	617278	Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive
DEPDC5	99.18 %	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant; Developmental and epileptic encephalopathy 111, 620504 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DES	100 %	125660	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Cardiomyopathy, dilated, 1I, 604765 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal dominant, Autosomal recessive
DGAT1	100 %	604900	Diarrhea 7, protein-losing enteropathy type, 615863 (3), Autosomal recessive
DGKE	99.1 %	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
DGUOK	99.93 %	601465	Portal hypertension, noncirrhotic, 1, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
DHCR24	99.93 %	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHDDS	98.65 %	608172	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHFR	98.89 %	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DHH	100 %	605423	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 (3), Autosomal recessive; 46XY sex reversal 7, 233420 (3), Autosomal recessive
DHODH	99.99 %	126064	Miller syndrome, 263750 (3), Autosomal recessive
DHPS	93.17 %	600944	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive
DHTKD1	99.95 %	614984	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive
DHX16	99.98 %	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
DHX30	99.96 %	616423	Neurodevelopmental disorder with variable motor and speech impairment, 617804 (3), Autosomal dominant
DHX37	99.98 %	617362	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46XY sex reversal 11, 273250 (3), Autosomal dominant
DHX38	99.97 %	605584	Retinitis pigmentosa 84, 618220 (3), Autosomal recessive
DIABLO	100 %	605219	Deafness, autosomal dominant 64, 614152 (3), Autosomal dominant
DIAPH1	99.95 %	602121	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DIAPH2	99.05 %	300108	?Premature ovarian failure 2A, 300511 (3), X-linked dominant
DIAPH3	99.85 %	614567	Auditory neuropathy, autosomal dominant 1, 609129 (3), Autosomal dominant
DICER1	99.96 %	606241	Pleuropulmonary blastoma, 601200 (3), Autosomal dominant; Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3), Autosomal dominant; GLOW syndrome, somatic mosaic, 618272 (3); Rhabdomyosarcoma, embryonal, 2, 180295 (3)
DIO1	99.99 %	147892	Thyroid hormone metabolism, abnormal, 2, 619855 (3), Autosomal dominant
DIP2B	99.65 %	611379	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630 (3), Autosomal dominant
DIS3L2	99.9 %	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DISC1	96.3 %	605210	{Schizophrenia 9, susceptibility to}, 604906 (3)
DISP1	99.93 %	607502	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DKC1	99.59 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLAT	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLC1	99.98 %	604258	Colorectal cancer, somatic, 114500 (3)
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DLG3	99.96 %	300189	Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive
DLG4	99.99 %	602887	Intellectual developmental disorder, autosomal dominant 62, 618793 (3), Autosomal dominant
DLG5	99.91 %	604090	Yuksel-Vogel-Bauser syndrome, 620703 (3), Autosomal recessive
DLL1	99.99 %	606582	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant
DLL3	100 %	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
DLL4	100 %	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DLST	99.99 %	126063	Pheochromocytoma/paraganglioma syndrome 7, 618475 (3), Autosomal dominant
DLX3	99.98 %	600525	Trichodontoosseous syndrome, 190320 (3), Autosomal dominant; Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant
DLX4	99.97 %	601911	?Orofacial cleft 15, 616788 (3), Autosomal dominant
DLX5	99.98 %	600028	Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
DMD	99.76 %	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DMGDH	99.95 %	605849	Dimethylglycine dehydrogenase deficiency, 605850 (3), Autosomal recessive
DMP1	99.99 %	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DMPK	99.93 %	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
DMXL2	99.86 %	612186	Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
DNA2	99.83 %	601810	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 (3), Autosomal dominant; Rothmund-Thomson syndrome, type 4, 620819 (3), Autosomal recessive; Seckel syndrome 8, 615807 (3), Autosomal recessive
DNAAF1	99.99 %	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF11	99.8 %	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
DNAAF2	99.91 %	612517	Ciliary dyskinesia, primary, 10, 612518 (3), Autosomal recessive
DNAAF3	99.99 %	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
DNAAF4	99.78 %	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive
DNAAF5	99.99 %	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
DNAAF6	98.73 %	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
DNAH1	99.98 %	603332	Spermatogenic failure 18, 617576 (3), Autosomal recessive; Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive
DNAH10	99.93 %	605884	Spermatogenic failure 56, 619515 (3), Autosomal recessive
DNAH11	99.93 %	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH17	99.98 %	610063	Spermatogenic failure 39, 618643 (3), Autosomal recessive
DNAH2	99.33 %	603333	Spermatogenic failure 45, 619094 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DNAH5	99.98 %	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
DNAH7	99.66 %	610061	Ciliary dyskinesia, primary, 50, 620356 (3), Autosomal recessive
DNAH8	99.84 %	603337	Spermatogenic failure 46, 619095 (3), Autosomal recessive
DNAH9	99.99 %	603330	Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive
DNAI1	99.92 %	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	99.86 %	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive
DNAJB11	99.97 %	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
DNAJB13	99.91 %	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
DNAJB2	99.97 %	604139	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881 (3), Autosomal recessive
DNAJB4	99.06 %	611327	Congenital myopathy 21 with early respiratory failure, 620326 (3), Autosomal recessive
DNAJB6	99.97 %	611332	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511 (3), Autosomal dominant
DNAJC12	99.72 %	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
DNAJC19	99.76 %	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC21	99.67 %	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
DNAJC3	99.92 %	601184	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
DNAJC30	100 %	618202	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382 (3), Autosomal recessive
DNAJC5	99.99 %	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant
DNAJC6	99.48 %	608375	Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive
DNAL1	99.79 %	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
DNAL4	99.89 %	610565	?Mirror movements 3, 616059 (3), Autosomal recessive
DNALI1	99.99 %	602135	Spermatogenic failure 83, 620354 (3), Autosomal recessive
DNASE1	100 %	125505	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
DNASE1L3	99.9 %	602244	Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive
DNASE2	100 %	126350	Autoinflammatory-pancytopenia syndrome, 619858 (3), Autosomal recessive
DNHD1	100 %	617277	Spermatogenic failure 65, 619712 (3), Autosomal recessive
DNM1	92.28 %	602377	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352 (3), Autosomal recessive; Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346 (3), Autosomal dominant
DNM1L	99.4 %	603850	Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal dominant, Autosomal recessive
DNM2	99.99 %	602378	Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive
DNMBP	99.94 %	611282	Cataract 48, 618415 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DNMT1	99.13 %	126375	Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant; Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant
DNMT3A	100 %	602769	Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant
DNMT3B	99.98 %	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
DOCK11	99.45 %	300681	Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109 (3), X-linked recessive
DOCK2	100 %	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
DOCK3	99.96 %	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive
DOCK6	100 %	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK7	94.41 %	615730	Developmental and epileptic encephalopathy 23, 615859 (3), Autosomal recessive
DOCK8	99.86 %	611432	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700 (3), Autosomal recessive
DOHH	100 %	611262	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066 (3), Autosomal recessive
DOK7	99.97 %	610285	Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
DOLK	100 %	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DONSON	99.99 %	611428	Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive
DPAGT1	100 %	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive
DPF2	99.99 %	601671	Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant
DPH1	100 %	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive
DPH2	99.97 %	603456	Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062 (3), Autosomal recessive
DPH5	96.83 %	611075	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070 (3), Autosomal recessive
DPM1	90.68 %	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPM2	100 %	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
DPM3	99.98 %	605951	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive
DPP6	99.99 %	126141	Intellectual developmental disorder, autosomal dominant 33, 616311 (3), Autosomal dominant; {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
DPP9	100 %	608258	Hatipoglu immunodeficiency syndrome, 620331 (3), Autosomal recessive
DPY19L2	92.02 %	613893	Spermatogenic failure 9, 613958 (3), Autosomal recessive
DPYD	94.53 %	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
DPYS	99.99 %	613326	Dihydropyrimidinuria, 222748 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DPYSL5	99.93 %	608383	Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant
DRAM2	96.06 %	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
DRC1	99.93 %	615288	Spermatogenic failure 80, 620222 (3), Autosomal recessive; Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
DRD3	100 %	126451	{Essential tremor, hereditary, 1}, 190300 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
DRD4	99.99 %	126452	{Attention deficit-hyperactivity disorder}, 143465 (3), Autosomal dominant; Autonomic nervous system dysfunction (3)
DRD5	100 %	126453	{Blepharospasm, primary benign}, 606798 (3), Autosomal dominant; {Attention deficit-hyperactivity disorder, susceptibility to}, 143465 (3), Autosomal dominant
DRG1	99.95 %	603952	Tan-Almurshedi syndrome, 620641 (3), Autosomal recessive
DSC2	99.73 %	125645	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal dominant, Autosomal recessive; Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal dominant, Autosomal recessive
DSC3	99.79 %	600271	Hypotrichosis and recurrent skin vesicles, 613102 (3), Autosomal recessive
DSE	100 %	605942	Ehlers-Danlos syndrome, musculocontractural type 2, 615539 (3), Autosomal recessive
DSG1	99.35 %	125670	Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive
DSG2	99.96 %	125671	Cardiomyopathy, dilated, 1BB, 612877 (3), Autosomal recessive; Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant
DSG3	99.97 %	169615	Blistering, acantholytic, of oral and laryngeal mucosa, 619226 (3), Autosomal recessive
DSG4	99.91 %	607892	Hypotrichosis 6, 607903 (3), Autosomal recessive
DSP	100 %	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive
DSPP	99.99 %	125485	Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant
DST	99.52 %	113810	Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive; Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425 (3), Autosomal recessive
DSTYK	99.83 %	612666	Spastic paraplegia 23, autosomal recessive, 270750 (3), Autosomal recessive; Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant
DTNA	100 %	601239	Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3), Autosomal dominant
DTNBP1	99.89 %	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
DTYMK	99.99 %	188345	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 (3), Autosomal recessive
DUOX2	98.85 %	606759	Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive
DUOXA2	100 %	612772	Thyroid dyshormonogenesis 5, 274900 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DUSP6	99.99 %	602748	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant
DUT	99.9 %	601266	Bone marrow failure and diabetes mellitus syndrome, 620044 (3), Autosomal recessive
DVL1	100 %	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
DVL3	99.99 %	601368	Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant
DYM	99.96 %	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
DYNC1H1	99.99 %	600112	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant
DYNC1I2	98.97 %	603331	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (3), Autosomal recessive
DYNC2H1	99.66 %	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Digenic recessive, Autosomal recessive
DYNC2I1	99.99 %	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
DYNC2I2	99.98 %	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
DYNC2LI1	99.94 %	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
DYNLT2B	100 %	617353	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive
DYRK1A	99.98 %	600855	Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant
DYRK1B	99.99 %	604556	Abdominal obesity-metabolic syndrome 3, 615812 (3), Autosomal dominant
DYSF	99.95 %	603009	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
DZIP1	99.97 %	608671	Spermatogenic failure 47, 619102 (3), Autosomal recessive; ?Mitral valve prolapse 3, 610840 (3), Autosomal dominant
DZIP1L	98.46 %	617570	Polycystic kidney disease 5, 617610 (3), Autosomal recessive
EARS2	99.96 %	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
EBF3	99.99 %	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
EBP	99.92 %	300205	MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant
ECE1	99.92 %	600423	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; ?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 (3), Autosomal dominant
ECEL1	100 %	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
ECHS1	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
ECM1	99.99 %	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
EDA	99.44 %	300451	Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EDAR	99.89 %	604095	[Hair morphology 1, hair thickness], 612630 (3); Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive
EDARADD	99.98 %	606603	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive; Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant
EDC3	99.97 %	609842	?Intellectual developmental disorder, autosomal recessive 50, 616460 (3), Autosomal recessive
EDEM3	98.38 %	610214	Congenital disorder of glycosylation, type IIv, 619493 (3), Autosomal recessive
EDN1	99.99 %	131240	Question mark ears, isolated, 612798 (3), Autosomal dominant; Auriculocondylar syndrome 3, 615706 (3), Autosomal recessive
EDN3	100 %	131242	Waardenburg syndrome, type 4B, 613265 (3), Autosomal dominant, Autosomal recessive; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant
EDNRA	99.97 %	131243	{Migraine, resistance to}, 157300 (3), Autosomal dominant; Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant
EDNRB	99.99 %	131244	{Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; ?ABCD syndrome, 600501 (3), Autosomal recessive; Waardenburg syndrome, type 4A, 277580 (3), Autosomal dominant, Autosomal recessive
EED	93.81 %	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant
EEF1A2	100 %	602959	Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 38, 616393 (3), Autosomal dominant
EEF2	99.95 %	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
EFCAB1	99.96 %	619564	Ciliary dyskinesia, primary, 53, 620642 (3), Autosomal recessive
EFEMP1	99.68 %	601548	Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type ID, 620780 (3), Autosomal recessive; Glaucoma 1, open angle, H, 611276 (3), Autosomal dominant
EFEMP2	99.94 %	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
EFHC1	99.99 %	608815	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631 (3), Autosomal dominant; {Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 (3), Autosomal dominant
EFL1	99.83 %	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
EFNB1	99.95 %	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
EFTUD2	99.93 %	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EGF	99.96 %	131530	?Hypomagnesemia 4, renal, 611718 (3), Autosomal recessive
EGFR	99.68 %	131550	Neonatal nephrocutaneous inflammatory syndrome, 616069 (3), Autosomal recessive; Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Somatic mutation, Autosomal dominant; Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Somatic mutation, Autosomal dominant; {Nonsmall cell lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
EGLN1	99.86 %	606425	Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant; [Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant
EGR2	100 %	129010	Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal dominant, Autosomal recessive
EHBP1	99.73 %	609922	{Prostate cancer, hereditary, 12}, 611868 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EHBP1L1	100 %	619583	*No OMIM phenotypes*
EHHADH	99.99 %	607037	?Fanconi renotubular syndrome 3, 615605 (3), Autosomal dominant
EHMT1	98.38 %	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
EIF2AK1	99.9 %	613635	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3), Autosomal dominant
EIF2AK2	99.7 %	176871	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant; Dystonia 33, 619687 (3), Autosomal dominant, Autosomal recessive
EIF2AK3	97.43 %	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
EIF2AK4	99.97 %	609280	Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive
EIF2B1	99.98 %	606686	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive
EIF2B2	99.9 %	606454	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3), Autosomal recessive
EIF2B3	97.26 %	606273	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3), Autosomal recessive
EIF2B4	99.96 %	606687	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3), Autosomal recessive
EIF2B5	99.98 %	603945	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3), Autosomal recessive
EIF2S3	99.34 %	300161	MEHMO syndrome, 300148 (3), X-linked recessive
EIF3F	99.96 %	603914	Intellectual developmental disorder, autosomal recessive 67, 618295 (3), Autosomal recessive
EIF4A2	99.99 %	601102	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455 (3), Autosomal dominant, Autosomal recessive
EIF4A3	99.99 %	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
EIF4E	89.46 %	133440	{Autism, susceptibility to, 19}, 615091 (3)
EIF4G1	100 %	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
EIF5A	100 %	600187	Faundes-Banka syndrome, 619376 (3), Autosomal dominant
ELAC2	99.9 %	605367	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive
ELANE	100 %	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
ELF4	99.97 %	300775	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074 (3), X-linked recessive
ELMO2	100 %	606421	Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive
ELMOD3	99.88 %	615427	?Deafness, autosomal recessive 88, 615429 (3), Autosomal recessive; ?Deafness, autosomal dominant 81, 619500 (3), Autosomal dominant
ELN	99.86 %	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
ELOVL1	100 %	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant, Autosomal recessive
ELOVL4	99.91 %	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 (3), Autosomal recessive
ELOVL5	99.87 %	611805	Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ELP1	99.96 %	603722	{Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal dominant, Autosomal recessive; Dysautonomia, familial, 223900 (3), Autosomal recessive
ELP2	99.82 %	616054	Intellectual developmental disorder, autosomal recessive 58, 617270 (3), Autosomal recessive
ELP4	93.66 %	606985	?Aniridia 2, 617141 (3), Autosomal dominant
EMC1	99.85 %	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
EMC10	99.96 %	614545	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 (3), Autosomal recessive
EMD	99.93 %	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
EMG1	100 %	611531	Bowen-Conradi syndrome, 211180 (3), Autosomal recessive
EMILIN1	99.99 %	130660	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080 (3), Autosomal dominant; Arterial tortuosity-bone fragility syndrome, 620908 (3), Autosomal recessive
EML1	99.99 %	602033	Band heterotopia, 600348 (3), Autosomal recessive
EMP2	100 %	602334	Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive
EMX2	100 %	600035	Schizencephaly, 269160 (3)
EN1	100 %	131290	?ENDOVE syndrome, limb-brain type, 619218 (3), Autosomal recessive
ENAM	100 %	606585	Amelogenesis imperfecta, type IC, 204650 (3), Autosomal recessive; Amelogenesis imperfecta, type IB, 104500 (3), Autosomal dominant
ENG	100 %	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
ENO3	100 %	131370	Glycogen storage disease XIII, 612932 (3), Autosomal recessive
ENPP1	99.88 %	173335	{Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant
ENTPD1	99.98 %	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
EOGT	99.09 %	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
EP300	99.97 %	602700	Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPAS1	99.99 %	603349	Erythrocytosis, familial, 4, 611783 (3), Autosomal dominant
EPB41	97.07 %	130500	Elliptocytosis-1, 611804 (3), Autosomal dominant, Autosomal recessive
EPB41L1	100 %	602879	?Intellectual developmental disorder, autosomal dominant 11, 614257 (3), Autosomal dominant
EPB42	100 %	177070	Spherocytosis, type 5, 612690 (3)
EPCAM	99.89 %	185535	Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive; Lynch syndrome 8, 613244 (3), Autosomal dominant
EPG5	99.95 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPHA10	99.68 %	611123	?Deafness, autosomal dominant 88, 620283 (3), Autosomal dominant
EPHA2	99.99 %	176946	Cataract 6, multiple types, 116600 (3), Autosomal dominant
EPHB2	99.86 %	600997	?Bleeding disorder, platelet-type, 22, 618462 (3), Autosomal recessive; {Prostate cancer/brain cancer susceptibility, somatic}, 603688 (3)
EPHB4	99.82 %	600011	Capillary malformation-arteriovenous malformation 2, 618196 (3), Autosomal dominant; Lymphatic malformation 7, 617300 (3), Autosomal dominant
EPHX2	100 %	132811	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890 (3), Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EPM2A	99.99 %	607566	Myoclonic epilepsy of Lafora 1, 254780 (3), Autosomal recessive
EPO	99.89 %	133170	{Microvascular complications of diabetes 2}, 612623 (3); Erythrocytosis, familial, 5, 617907 (3), Autosomal dominant; ?Diamond-Blackfan anemia-like, 617911 (3), Autosomal recessive
EPOR	100 %	133171	[Erythrocytosis, familial, 1], 133100 (3), Autosomal dominant
EPRS1	99.53 %	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
EPS8	99.64 %	600206	?Deafness, autosomal recessive 102, 615974 (3), Autosomal recessive
EPS8L2	99.99 %	614988	Deafness autosomal recessive 106, 617637 (3), Autosomal recessive
EPS8L3	99.84 %	614989	?Hypotrichosis 5, 612841 (3), Autosomal dominant
EPX	99.99 %	131399	[Eosinophil peroxidase deficiency], 261500 (3), Autosomal recessive
ERAL1	99.99 %	607435	Perrault syndrome 6, 617565 (3), Autosomal recessive
ERBB2	99.98 %	164870	Gastric cancer, somatic, 613659 (3); Adenocarcinoma of lung, somatic, 211980 (3); Ovarian cancer, somatic, 167000 (3); ?Visceral neuropathy, familial, 2, autosomal recessive, 619465 (3), Autosomal recessive; Glioblastoma, somatic, 137800 (3)
ERBB3	99.91 %	190151	?Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive; {?Erythroleukemia, familial, susceptibility to}, 133180 (3), Autosomal dominant; Visceral neuropathy, familial, 1, autosomal recessive, 243180 (3), Autosomal recessive
ERBB4	99.92 %	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant
ERCC1	99.96 %	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC2	99.98 %	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
ERCC3	99.9 %	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC4	99.92 %	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive
ERCC5	99.99 %	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	99.6 %	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
ERCC6L2	99.94 %	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
ERCC8	99.79 %	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
ERF	99.98 %	611888	Craniosynostosis 4, 600775 (3), Autosomal dominant; Chitayat syndrome, 617180 (3), Autosomal dominant
ERG	100 %	165080	Lymphatic malformation 14, 620602 (3), Autosomal dominant
ERGIC1	100 %	617946	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100 (3), Autosomal recessive
ERI1	99.8 %	608739	Hoxha-Aliu syndrome, 620662 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ERLIN1	99.98 %	611604	Spastic paraplegia 62, autosomal recessive, 615681 (3), Autosomal recessive
ERLIN2	99.94 %	611605	Spastic paraplegia 18A, autosomal dominant, 620512 (3), Autosomal dominant; Spastic paraplegia 18B, autosomal recessive, 611225 (3), Autosomal recessive
ERMAP	99.76 %	609017	[Blood group, Scianna system], 111750 (3); [Blood group, Radin], 111620 (3)
ERMARD	99.94 %	615532	?Periventricular nodular heterotopia 6, 615544 (3), Autosomal dominant
ESAM	99.99 %	614281	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371 (3), Autosomal recessive
ESCO2	99.92 %	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
ESPN	100 %	606351	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 (3), Autosomal recessive; Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; ?Usher syndrome, type 1M, 618632 (3), Autosomal recessive
ESR1	100 %	133430	Breast cancer, somatic, 114480 (3); {Migraine, susceptibility to}, 157300 (3), Autosomal dominant; Estrogen resistance, 615363 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3)
ESR2	99.96 %	601663	?Ovarian dysgenesis 8, 618187 (3), Autosomal dominant
ESRP1	99.91 %	612959	?Deafness, autosomal recessive 109, 618013 (3), Autosomal recessive
ESRRB	100 %	602167	Deafness, autosomal recessive 35, 608565 (3), Autosomal recessive
ETFA	99.88 %	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	100 %	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	99.82 %	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
ETHE1	84.97 %	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
ETV6	99.99 %	600618	Thrombocytopenia 5, 616216 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3)
EVC	99.95 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.97 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EWSR1	99.94 %	133450	Neuroepithelioma, 612219 (3); Ewing sarcoma, 612219 (3)
EXOC2	99.97 %	615329	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive
EXOC3L2	99.88 %	616927	*No OMIM phenotypes*
EXOC6B	99.41 %	607880	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395 (3), Autosomal recessive
EXOC7	100 %	608163	Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3), Autosomal recessive
EXOC8	99.99 %	615283	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076 (3), Autosomal recessive
EXOSC1	100 %	606493	?Pontocerebellar hypoplasia, type 1F, 619304 (3), Autosomal recessive
EXOSC2	100 %	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
EXOSC3	100 %	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC5	99.98 %	606492	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 (3), Autosomal recessive
EXOSC8	99.91 %	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXOSC9	94.91 %	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EXPH5	99.81 %	612878	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028 (3), Autosomal recessive
EXT1	99.94 %	608177	Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation
EXT2	99.98 %	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
EXTL3	99.99 %	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
EYA1	99.81 %	601653	Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
EYA4	99.96 %	603550	?Cardiomyopathy, dilated, 1J, 605362 (3), Autosomal dominant; Deafness, autosomal dominant 10, 601316 (3), Autosomal dominant
EYS	99.86 %	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
EZH2	99.89 %	601573	Weaver syndrome, 277590 (3), Autosomal dominant
F10	99.98 %	613872	Factor X deficiency, 227600 (3), Autosomal recessive
F11	100 %	264900	Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3)
F12	99.99 %	610619	Angioedema, hereditary, 3, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
F13A1	99.43 %	134570	Factor XIIIA deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant
F13B	98.95 %	134580	Factor XIIIB deficiency, 613235 (3), Autosomal recessive
F2	99.99 %	176930	Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; Dysprothrombinemia, 613679 (3), Autosomal recessive; Thrombophilia 1 due to thrombin defect, 188050 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial
F5	99.59 %	612309	Thrombophilia 2 due to activated protein C resistance, 188055 (3), Autosomal dominant; {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant; {Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Factor V deficiency, 227400 (3), Autosomal recessive
F7	100 %	613878	{Myocardial infarction, decreased susceptibility to}, 608446 (3); Factor VII deficiency, 227500 (3), Autosomal recessive
F8	99.91 %	300841	Thrombophilia 13, X-linked, due to factor VIII defect, 301071 (3); Hemophilia A, 306700 (3), X-linked recessive
F9	99.87 %	300746	{Deep venous thrombosis, protection against}, 300807 (3), X-linked recessive; Hemophilia B, 306900 (3), X-linked recessive; Thrombophilia 8, X-linked, due to factor IX defect, 300807 (3), X-linked recessive; {Warfarin sensitivity}, 301052 (3), X-linked
FA2H	99.98 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAAH	99.87 %	602935	{Drug addiction, susceptibility to}, 606581 (3)
FADD	99.97 %	602457	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759 (3), Autosomal recessive
FAH	99.98 %	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FAM111A	100 %	615292	Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FAM111B	99.98 %	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FAM126A	99.81 %	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FAM149B1	99.69 %	618413	Joubert syndrome 36, 618763 (3), Autosomal recessive
FAM161A	99.77 %	613596	Retinitis pigmentosa 28, 606068 (3), Autosomal recessive
FAM20A	100 %	611062	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive
FAM20C	100 %	611061	Raine syndrome, 259775 (3), Autosomal recessive
FAM50A	99.99 %	300453	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261 (3), X-linked recessive
FAM83H	100 %	611927	Amelogenesis imperfecta, type IIIA, 130900 (3), Autosomal dominant
FAN1	99.73 %	613534	Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive
FANCA	100 %	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
FANCB	99.24 %	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
FANCC	99.98 %	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
FANCD2	99.86 %	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FANCE	99.99 %	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
FANCF	100 %	613897	Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive
FANCG	100 %	602956	Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive
FANCI	99.96 %	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
FANCL	99.67 %	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
FANCM	99.83 %	609644	Premature ovarian failure 15, 618096 (3), Autosomal recessive; Spermatogenic failure 28, 618086 (3), Autosomal recessive
FAR1	99.82 %	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
FARS2	100 %	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FARSA	100 %	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	99.64 %	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
FAS	99.99 %	134637	Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant
FASLG	99.84 %	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
FASTKD2	99.93 %	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
FAT2	99.99 %	604269	Spinocerebellar ataxia 45, 617769 (3), Autosomal dominant
FAT4	99.98 %	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
FBLN1	98.69 %	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant
FBLN5	100 %	604580	Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 (3), Autosomal dominant; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FBN1	99.85 %	134797	Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
FBN2	99.9 %	612570	Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant
FBP1	100 %	611570	Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive
FBP2	99.99 %	603027	?Leukodystrophy, childhood-onset, remitting, 619864 (3), Autosomal dominant
FBXL3	99.48 %	605653	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive
FBXL4	100 %	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
FBXO11	99.53 %	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant
FBXO28	99.67 %	609100	Developmental and epileptic encephalopathy 100, 619777 (3), Autosomal dominant
FBXO31	99.99 %	609102	?Intellectual developmental disorder, autosomal recessive 45, 615979 (3), Autosomal recessive
FBXO38	99.96 %	608533	Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575 (3), Autosomal dominant
FBXO43	99.96 %	609110	Spermatogenic failure 64, 619696 (3), Autosomal recessive; Oocyte/zygote/embryo maturation arrest 12, 619697 (3), Autosomal recessive
FBXO7	99.98 %	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
FBXW11	99.96 %	605651	Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant
FBXW7	99.9 %	606278	Developmental delay, hypotonia, and impaired language, 620012 (3), Autosomal dominant
FCGR1A	99.77 %	146760	[IgG receptor I, phagocytic, familial deficiency of] (3)
FCGR2A	99.99 %	146790	{Malaria, severe, susceptibility to}, 611162 (3); {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 (3), Autosomal recessive; {Lupus nephritis, susceptibility to}, 152700 (3), Autosomal dominant
FCGR2B	70.97 %	604590	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3)
FCGR3A	99.93 %	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
FCHO1	99.99 %	613437	Immunodeficiency 76, 619164 (3), Autosomal recessive
FCN3	99.48 %	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
FCSK	99.97 %	608675	Congenital disorder of glycosylation with defective fucosylation 2, 618324 (3), Autosomal recessive
FDFT1	99.99 %	184420	Squalene synthase deficiency, 618156 (3), Autosomal recessive
FDPS	100 %	134629	Porokeratosis 9, multiple types, 616631 (3), Autosomal dominant
FDX2	99.99 %	614585	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive
FDXR	99.99 %	103270	Multiple mitochondrial dysfunctions syndrome 9B, 620887 (3); Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive
FECH	99.98 %	612386	Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive
FERMT1	99.9 %	607900	Kindler syndrome, 173650 (3), Autosomal recessive
FERMT3	99.99 %	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FEZF1	100 %	613301	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive
FFAR4	100 %	609044	{Obesity, susceptibility to}, 607514 (3)
FGA	99.98 %	134820	Amyloidosis, hereditary systemic 2, 105200 (3), Autosomal dominant; Hypodysfibrinogenemia, congenital, 616004 (3); Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGB	99.93 %	134830	Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGD1	99.96 %	300546	Intellectual developmental disorder, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive
FGD4	99.87 %	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
FGF10	99.95 %	602115	LADD syndrome 3, 620193 (3), Autosomal dominant; Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant
FGF12	99.94 %	601513	Developmental and epileptic encephalopathy 47, 617166 (3), Autosomal dominant
FGF13	99.76 %	300070	Developmental and epileptic encephalopathy 90, 301058 (3), X-linked recessive, X-linked dominant; Intellectual developmental disorder, X-linked 110, 301095 (3), X-linked recessive
FGF14	99.99 %	601515	Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant; Spinocerebellar ataxia 27B, late-onset, 620174 (3), Autosomal dominant
FGF16	99.6 %	300827	Metacarpal 4-5 fusion, 309630 (3), X-linked recessive
FGF17	99.81 %	603725	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant
FGF20	99.71 %	605558	?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive
FGF23	100 %	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant
FGF3	99.97 %	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
FGF5	99.97 %	165190	Trichomegaly, 190330 (3), Autosomal recessive
FGF8	100 %	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGF9	100 %	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant
FGFR1	100 %	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR2	99.99 %	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FGFR3	100 %	134934	Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal dominant, Autosomal recessive; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); LADD syndrome 2, 620192 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant
FGFR4	100 %	134935	{Cancer progression/metastasis} (3)
FGG	99.98 %	134850	Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FH	99.95 %	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive
FHL1	99.97 %	300163	Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant
FHOD3	99.99 %	609691	Cardiomyopathy, familial hypertrophic, 28, 619402 (3), Autosomal dominant
FIBP	99.83 %	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FICD	100 %	620875	Spastic paraplegia 92, autosomal recessive, 620911 (3), Autosomal recessive
FIG4	99.83 %	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive
FIGLA	99.95 %	608697	Premature ovarian failure 6, 612310 (3), Autosomal dominant, Autosomal recessive
FILIP1	99.98 %	607307	Neuromuscular disorder, congenital, with dysmorphic facies, 620775 (3), Autosomal recessive
FITM2	99.99 %	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive
FKBP10	99.98 %	607063	Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive; Bruck syndrome 1, 259450 (3), Autosomal recessive
FKBP14	99.97 %	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
FKBP5	99.96 %	602623	{Major depressive disorder and accelerated response to antidepressant drug treatment}, 608516 (3)
FKBP6	98.73 %	604839	Spermatogenic failure 77, 620103 (3), Autosomal recessive
FKRP	100 %	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FKTN	99.94 %	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive
FLAD1	99.98 %	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive
FLCN	99.51 %	607273	Birt-Hogg-Dube syndrome, 135150 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Pneumothorax, primary spontaneous, 173600 (3), Autosomal dominant; Renal carcinoma, chromophobe, somatic, 144700 (3)
FLG	98.88 %	135940	Ichthyosis vulgaris, 146700 (3), Autosomal dominant, Autosomal recessive; {Dermatitis, atopic, susceptibility to, 2}, 605803 (3)
FLG2	99.84 %	616284	Peeling skin syndrome 6, 618084 (3), Autosomal recessive
FLI1	100 %	193067	Bleeding disorder, platelet-type, 21, 617443 (3), Autosomal dominant, Autosomal recessive
FLII	99.95 %	600362	Cardiomyopathy, dilated, 2J, 620635 (3), Autosomal recessive
FLNA	99.99 %	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FLNB	99.98 %	603381	Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Boomerang dysplasia, 112310 (3), Autosomal dominant
FLNC	99.99 %	102565	Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia, familial, 617047 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant
FLRT3	100 %	604808	Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant
FLT3	99.93 %	136351	Leukemia, acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, reduced survival in, somatic, 601626 (3); Leukemia, acute myeloid, somatic, 601626 (3)
FLT3LG	99.91 %	600007	?Immunodeficiency 125, 620926 (3), Autosomal recessive
FLT4	97.93 %	136352	Hemangioma, capillary infantile, somatic, 602089 (3); Lymphatic malformation 1, 153100 (3), Autosomal dominant; Congenital heart defects, multiple types, 7, 618780 (3), Autosomal dominant
FLVCR1	99.91 %	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FLVCR2	100 %	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
FMN2	99.99 %	606373	Intellectual developmental disorder, autosomal recessive 47, 616193 (3), Autosomal recessive
FMO3	99.93 %	136132	Trimethylaminuria, 602079 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FMR1	99.56 %	309550	Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
FN1	99.95 %	135600	Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant; Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant
FNIP1	99.87 %	610594	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 (3), Autosomal recessive
FOCAD	99.62 %	614606	Liver disease, severe congenital, 619991 (3), Autosomal recessive
FOLR1	100 %	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FOSL2	99.95 %	601575	Aplasia cutis-enamel dysplasia syndrome, 620789 (3), Autosomal dominant
FOXC1	100 %	601090	Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant
FOXC2	100 %	602402	Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant
FOXD3	99.85 %	611539	{Autoimmune disease, susceptibility to, 1}, 607836 (3), Autosomal dominant
FOXE1	99.95 %	602617	Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant
FOXE3	99.29 %	601094	Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3)
FOXF1	99.99 %	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXG1	99.91 %	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXI1	100 %	601093	Enlarged vestibular aqueduct, 600791 (3), Autosomal recessive
FOXI3	100 %	612351	Craniofacial microsomia 2, 620444 (3), Autosomal dominant, Autosomal recessive
FOXJ1	100 %	602291	Ciliary dyskinesia, primary, 43, 618699 (3), Autosomal dominant
FOXL1	100 %	603252	Otosclerosis 11, 620576 (3), Autosomal dominant
FOXL2	99.97 %	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal dominant, Autosomal recessive; Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal dominant, Autosomal recessive; Premature ovarian failure 3, 608996 (3), Autosomal dominant
FOXN1	99.97 %	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3), Autosomal dominant; T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
FOXO1	100 %	136533	Rhabdomyosarcoma, alveolar, 268220 (3), Somatic mutation
FOXP1	99.98 %	605515	Intellectual developmental disorder with language impairment with or without autistic features, 613670 (3), Autosomal dominant
FOXP2	99.98 %	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
FOXP3	99.93 %	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
FOXRED1	100 %	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
FRA10AC1	99.76 %	608866	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113 (3), Autosomal recessive
FRAS1	99.97 %	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FREM1	99.98 %	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
FREM2	99.97 %	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
FRMD4A	100 %	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMD5	100 %	616309	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 (3), Autosomal dominant
FRMD7	99.97 %	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
FRMPD4	99.92 %	300838	Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked
FRRS1L	99.91 %	604574	Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive
FRZB	99.99 %	605083	{Osteoarthritis susceptibility 1}, 165720 (3), Multifactorial
FSCN2	100 %	607643	Retinitis pigmentosa 30, 607921 (3)
FSHB	100 %	136530	Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive
FSHR	99.99 %	136435	Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian dysgenesis 1, 233300 (3), Autosomal recessive
FSIP2	99.92 %	615796	Spermatogenic failure 34, 618153 (3), Autosomal recessive
FTCD	99.99 %	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
FTH1	22.62 %	134770	Neurodegeneration with brain iron accumulation 9, 620669 (3), Autosomal dominant; ?Hemochromatosis, type 5, 615517 (3), Autosomal dominant
FTL	99.99 %	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal dominant, Autosomal recessive; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
FTO	99.66 %	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
FTSJ1	99.95 %	300499	Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive
FUCA1	98.72 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUS	99.93 %	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
FUT1	100 %	211100	[Bombay phenotype], 616754 (3), Autosomal recessive
FUT2	100 %	182100	{Norwalk virus infection, resistance to} (3); {Vitamin B12 plasma level QTL1}, 612542 (3); [Bombay phenotype, digenic], 616754 (3), Autosomal recessive
FUT3	100 %	111100	[Blood group, Lewis], 618983 (3)
FUT6	100 %	136836	[Fucosyltransferase 6 deficiency], 613852 (3)
FUT8	99.98 %	602589	Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive
FUZ	99.99 %	610622	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
FXN	99.96 %	606829	Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive; Friedreich ataxia, 229300 (3), Autosomal recessive
FXR1	99.82 %	600819	Congenital myopathy 9B, proximal, with minicore lesions, 618823 (3), Autosomal recessive; ?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822 (3), Autosomal recessive
FXYD2	100 %	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FYB1	99.95 %	602731	Thrombocytopenia 3, 273900 (3), Autosomal recessive
FYCO1	100 %	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
FZD2	99.98 %	600667	Omodysplasia 2, 164745 (3), Autosomal dominant
FZD4	100 %	604579	Retinopathy of prematurity, 133780 (3), Autosomal dominant; Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant
FZD5	100 %	601723	Microphthalmia/coloboma 11, 620731 (3), Autosomal dominant
FZD6	99.95 %	603409	Nail disorder, nonsyndromic congenital, 1, 161050 (3), Autosomal recessive
FZR1	99.99 %	603619	Developmental and epileptic encephalopathy 109, 620145 (3), Autosomal dominant
G6PC1	99.93 %	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
G6PC3	99.98 %	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
G6PD	99.97 %	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
GAA	100 %	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
GAB1	99.89 %	604439	?Deafness, autosomal recessive 26, 605428 (3), Autosomal recessive
GABBR1	99.91 %	603540	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502 (3), Autosomal dominant
GABBR2	99.96 %	607340	{Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3); Developmental and epileptic encephalopathy 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant
GABRA1	100 %	137160	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3)
GABRA2	99.9 %	137140	Developmental and epileptic encephalopathy 78, 618557 (3), Autosomal dominant; {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial
GABRA3	99.96 %	305660	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091 (3), X-linked
GABRA5	99.96 %	137142	Developmental and epileptic encephalopathy 79, 618559 (3), Autosomal dominant
GABRB1	99.97 %	137190	Developmental and epileptic encephalopathy 45, 617153 (3), Autosomal dominant
GABRB2	99.92 %	600232	Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant
GABRB3	99.66 %	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Developmental and epileptic encephalopathy 43, 617113 (3), Autosomal dominant
GABRD	99.99 %	137163	{?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant
GABRG2	91.88 %	137164	Developmental and epileptic encephalopathy 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 3, 607681 (3), Autosomal dominant
GAD1	99.92 %	605363	Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive
GAL	99.9 %	137035	?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant
GALC	99.92 %	606890	Krabbe disease, 245200 (3), Autosomal recessive
GALE	99.9 %	606953	Thrombocytopenia 13, syndromic, 620776 (3), Autosomal recessive; Galactose epimerase deficiency, 230350 (3), Autosomal recessive
GALK1	100 %	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
GALM	100 %	137030	Galactosemia IV, 618881 (3), Autosomal recessive
GALNS	99.98 %	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
GALNT12	99.95 %	610290	{Colorectal cancer, susceptibility to, 1}, 608812 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GALNT2	99.44 %	602274	Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive
GALNT3	99.52 %	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
GALT	100 %	606999	Galactosemia, 230400 (3), Autosomal recessive
GAMT	100 %	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GAN	99.98 %	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
GANAB	99.97 %	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
GARS1	99.93 %	600287	Spinal muscular atrophy, infantile, James type, 619042 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant
GAS2	99.98 %	602835	?Deafness, autosomal recessive 125, 620877 (3), Autosomal recessive
GAS2L2	100 %	611398	?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive
GAS8	100 %	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
GATA1	99.97 %	305371	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595 (3); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive; Hemolytic anemia due to elevated adenosine deaminase, 301083 (3), X-linked recessive
GATA2	99.99 %	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GATA3	99.96 %	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
GATA4	99.99 %	600576	Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant
GATA5	100 %	611496	Congenital heart defects, multiple types, 5, 617912 (3), Autosomal dominant, Autosomal recessive
GATA6	99.9 %	601656	Atrial septal defect 9, 614475 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
GATAD1	99.79 %	614518	?Cardiomyopathy, dilated, 2B, 614672 (3), Autosomal recessive
GATAD2B	99.44 %	614998	GAND syndrome, 615074 (3), Autosomal dominant
GATB	99.95 %	603645	?Combined oxidative phosphorylation deficiency 41, 618838 (3), Autosomal recessive
GATC	99.99 %	617210	Combined oxidative phosphorylation deficiency 42, 618839 (3), Autosomal recessive
GATM	99.92 %	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
GBA	96.92 %	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
GBA2	99.99 %	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GBE1	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GBF1	99.97 %	603698	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483 (3), Autosomal dominant
GCDH	100 %	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCGR	100 %	138033	Mahvash disease, 619290 (3), Autosomal recessive
GCH1	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCK	99.99 %	138079	MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant
GCKR	99.99 %	600842	[Fasting plasma glucose level QTL 5], 613463 (3)
GCLC	99.83 %	606857	{Myocardial infarction, susceptibility to}, 608446 (3); Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 (3), Autosomal recessive
GCLM	94.51 %	601176	{Myocardial infarction, susceptibility to}, 608446 (3)
GCM2	100 %	603716	Hypoparathyroidism, familial isolated 2, 618883 (3), Autosomal dominant, Autosomal recessive; Hyperparathyroidism 4, 617343 (3), Autosomal dominant
GCNA	99.32 %	300369	Spermatogenic failure, X-linked, 4, 301077 (3), X-linked
GCNT2	100 %	600429	[Blood group, Ii], 110800 (3), Autosomal dominant; Adult i phenotype without cataract, 110800 (3), Autosomal dominant; Cataract 13 with adult i phenotype, 116700 (3), Autosomal recessive
GCSH	98.75 %	238330	Multiple mitochondrial dysfunctions syndrome 7, 620423 (3), Autosomal recessive
GDAP1	99.98 %	606598	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive
GDAP2	95.02 %	618128	Spinocerebellar ataxia, autosomal recessive 27, 618369 (3), Autosomal recessive
GDF1	100 %	602880	Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive
GDF11	99.97 %	603936	?Vertebral hypersegmentation and orofacial anomalies, 619122 (3), Autosomal dominant
GDF15	100 %	605312	{Hyperemesis gravidarum, susceptibility to}, 620730 (3), Autosomal dominant
GDF2	100 %	605120	Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant
GDF3	100 %	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia, isolated, with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant
GDF5	100 %	601146	Acromesomelic dysplasia 2A, 200700 (3), Autosomal recessive; Acromesomelic dysplasia 2B, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type C, 113100 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type A1, C, 615072 (3), Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GDF6	100 %	601147	Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant
GDF9	99.98 %	601918	Premature ovarian failure 14, 618014 (3), Autosomal recessive
GDI1	99.99 %	300104	Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant
GDNF	99.99 %	600837	{Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant
GEMIN4	100 %	606969	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive
GEMIN5	99.96 %	607005	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive
GET3	99.99 %	601913	?Cardiomyopathy, dilated, 2H, 620203 (3), Autosomal recessive
GET4	99.99 %	612056	?Congenital disorder of glycosylation, type IIy, 620200 (3), Autosomal recessive
GFAP	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
GFER	100 %	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
GFI1	99.88 %	600871	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant
GFI1B	99.99 %	604383	Bleeding disorder, platelet-type, 17, 187900 (3), Autosomal dominant, Autosomal recessive
GFM1	99.95 %	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GFM2	99.87 %	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GFPT1	99.79 %	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
GFRA1	100 %	601496	Renal hypodysplasia/aplasia 4, 619887 (3), Autosomal recessive
GGCX	99.88 %	137167	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 (3), Autosomal recessive; Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (3)
GGN	99.99 %	609966	Spermatogenic failure 69, 619826 (3), Autosomal recessive
GGPS1	99.95 %	606982	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518 (3), Autosomal recessive
GGT1	100 %	612346	?Glutathioninuria, 231950 (3), Autosomal recessive
GGT2	50.08 %	137181	[Gamma-glutamyltransferase, familial high serum], 137181 (2)
GH1	100 %	139250	Kowarski syndrome, 262650 (3), Autosomal recessive; Growth hormone deficiency, isolated, type II, 173100 (3), Autosomal dominant; Growth hormone deficiency, isolated, type IB, 612781 (3); Growth hormone deficiency, isolated, type IA, 262400 (3), Autosomal recessive
GHR	99.53 %	600946	Laron dwarfism, 262500 (3), Autosomal recessive; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant; {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant, Autosomal recessive
GHRH	100 %	139190	Gigantism due to GHRF hypersecretion (1); ?Isolated growth hormone deficiency due to defect in GHRF (1)
GHRHR	100 %	139191	Growth hormone deficiency, isolated, type IV, 618157 (3), Autosomal recessive
GHRL	99.99 %	605353	{Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GHSR	100 %	601898	Growth hormone deficiency, isolated partial, 615925 (3), Autosomal dominant, Autosomal recessive
GIGYF2	99.94 %	612003	{Parkinson disease 11}, 607688 (3)
GIMAP5	100 %	608086	Portal hypertension, noncirrhotic, 2, 619463 (3), Autosomal recessive
GINS1	99.99 %	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
GIPC1	99.98 %	605072	Oculopharyngodistal myopathy 2, 618940 (3), Autosomal dominant
GIPC3	99.97 %	608792	Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GJA3	100 %	121015	Cataract 14, multiple types, 601885 (3), Autosomal dominant
GJA5	100 %	121013	Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (GJA5/SCN5A), 108770 (3), Autosomal dominant
GJA8	99.99 %	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
GJB1	100 %	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
GJB2	100 %	121011	Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Digenic dominant, Autosomal recessive; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant
GJB3	99.99 %	603324	Deafness, digenic, GJB2/GJB3, 220290 (3), Digenic dominant, Autosomal recessive; Erythrokeratodermia variabilis et progressiva 1, 133200 (3), Autosomal dominant, Autosomal recessive; Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644 (3), Autosomal dominant
GJB4	100 %	605425	Erythrokeratodermia variabilis et progressiva 2, 617524 (3), Autosomal dominant
GJB6	100 %	604418	Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant; Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Digenic dominant, Autosomal recessive
GJC2	100 %	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
GK	99.27 %	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
GLA	99.9 %	300644	Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked
GLB1	100 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLCCI1	99.7 %	614283	{Glucocorticoid therapy, response to}, 614400 (3)
GLDC	99.99 %	238300	Glycine encephalopathy1, 605899 (3), Autosomal recessive
GLDN	99.98 %	608603	Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GLE1	99.99 %	603371	Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive; Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive
GLI1	99.98 %	165220	Polydactyly, preaxial I, 174400 (3), Autosomal recessive; Polydactyly, postaxial, type A8, 618123 (3), Autosomal recessive
GLI2	99.93 %	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	100 %	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLIS2	100 %	608539	Nephronophthisis 7, 611498 (3), Autosomal recessive
GLIS3	99.99 %	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
GLMN	91.1 %	601749	Glomuvenous malformations, 138000 (3), Autosomal dominant
GLRA1	100 %	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
GLRA2	99.87 %	305990	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076 (3), X-linked
GLRB	99.79 %	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLRX5	100 %	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
GLS	99.78 %	138280	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive
GLUD1	99.8 %	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
GLUL	99.88 %	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive; Developmental and epileptic encephalopathy 116, 620806 (3), Autosomal dominant
GLYCTK	100 %	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive
GM2A	100 %	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMNN	99.73 %	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
GMPPA	99.97 %	615495	Alacrima, achalasia, and impaired intellectual development syndrome, 615510 (3), Autosomal recessive
GMPPB	100 %	615320	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive
GNA11	99.99 %	139313	Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant; Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant
GNAI1	99.7 %	139310	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854 (3), Autosomal dominant
GNAI2	99.98 %	139360	Ventricular tachycardia, idiopathic, 192605 (3), Autosomal dominant; Pituitary adenoma, ACTH-secreting, somatic (3)
GNAI3	98.51 %	139370	Auriculocondylar syndrome 1, 602483 (3), Autosomal dominant
GNAL	100 %	139312	Dystonia 25, 615073 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GNAO1	99.86 %	139311	Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNAQ	99.94 %	600998	Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3)
GNAS	100 %	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GNAS-AS1	0 %	610540	Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant
GNAT1	100 %	139330	Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive
GNAT2	99.87 %	139340	Achromatopsia 4, 613856 (3)
GNB1	100 %	139380	Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant
GNB2	99.99 %	139390	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 (3), Autosomal dominant; ?Sick sinus syndrome 4, 619464 (3), Autosomal dominant
GNB3	100 %	139130	Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
GNB4	99.96 %	610863	Charcot-Marie-Tooth disease, dominant intermediate F, 615185 (3), Autosomal dominant
GNB5	99.98 %	604447	Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173 (3), Autosomal recessive
GNE	99.99 %	603824	Sialuria, 269921 (3), Autosomal dominant; Thrombocytopenia 12 with or without myopathy, 620757 (3), Autosomal recessive; Nonaka myopathy, 605820 (3), Autosomal recessive
GNMT	100 %	606628	Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive
GNPAT	99.78 %	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPNAT1	99.99 %	616510	?Rhizomelic dysplasia, Ain-Naz type, 616510 (3)
GNPTAB	99.76 %	607840	Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive
GNPTG	100 %	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNRH1	99.96 %	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive
GNRHR	99.94 %	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
GNS	99.59 %	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GOLGA2	100 %	602580	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240 (3), Autosomal recessive
GON7	99.92 %	617436	Galloway-Mowat syndrome 9, 619603 (3), Autosomal recessive
GORAB	99.59 %	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GOSR2	98.92 %	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive
GOT1	100 %	138180	Aspartate aminotransferase, serum level of, QTL1, 614419 (3)
GOT2	99.78 %	138150	Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive
GP1BA	99.97 %	606672	Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive
GP1BB	99.93 %	138720	Giant platelet disorder, isolated, 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive
GP6	98.85 %	605546	Bleeding disorder, platelet-type, 11, 614201 (3), Autosomal recessive
GP9	100 %	173515	Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive
GPAA1	100 %	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
GPC3	99.6 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GPC4	99.89 %	300168	Keipert syndrome, 301026 (3), X-linked recessive
GPC6	99.98 %	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
GPD1	99.85 %	138420	Hypertriglyceridemia, transient infantile, 614480 (3), Autosomal recessive
GPD1L	99.97 %	611778	Brugada syndrome 2, 611777 (3), Autosomal dominant
GPD2	99.7 %	138430	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant
GPHN	99.94 %	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
GPI	99.99 %	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 (3), Autosomal recessive
GPIHBP1	100 %	612757	Hyperlipoproteinemia, type 1D, 615947 (3), Autosomal recessive
GPNMB	99.99 %	604368	Amyloidosis, primary localized cutaneous, 3, 617920 (3), Autosomal recessive
GPR101	100 %	300393	Pituitary adenoma 2, GH-secreting, 300943 (3), X-linked
GPR143	99.6 %	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
GPR156	99.98 %	610464	Deafness, autosomal recessive 121, 620551 (3), Autosomal recessive
GPR161	99.92 %	612250	{Medulloblastoma predisposition syndrome}, 155255 (3), Somatic mutation, Autosomal dominant, Autosomal recessive
GPR179	99.99 %	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
GPR68	100 %	601404	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 (3), Autosomal recessive
GPR88	100 %	607468	?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive
GPRASP2	100 %	300969	?Deafness, X-linked 7, 301018 (3), X-linked recessive
GPRC5B	99.81 %	605948	Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447 (3), Autosomal dominant
GPSM2	96.11 %	609245	Chudley-McCullough syndrome, 604213 (3), Autosomal recessive
GPT2	99.97 %	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
GPX1	99.71 %	138320	Hemolytic anemia due to glutathione peroxidase deficiency, 614164 (1), Autosomal recessive
GPX4	100 %	138322	Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GRAP	68.27 %	604330	Deafness, autosomal recessive 114, 618456 (3), Autosomal recessive
GREB1L	99.99 %	617782	Deafness, autosomal dominant 80, 619274 (3), Autosomal dominant; Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant
GREM2	100 %	608832	Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant
GRHL2	100 %	608576	Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant
GRHL3	100 %	608317	van der Woude syndrome 2, 606713 (3), Autosomal dominant
GRHPR	99.93 %	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
GRIA1	99.97 %	138248	?Intellectual developmental disorder, autosomal recessive 76, 619931 (3), Autosomal recessive; Intellectual developmental disorder, autosomal dominant 67, 619927 (3), Autosomal dominant
GRIA2	99.99 %	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant
GRIA3	99.93 %	305915	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive
GRIA4	99.93 %	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant
GRID2	99.97 %	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRIK2	99.9 %	138244	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 (3), Autosomal dominant; Intellectual developmental disorder, autosomal recessive 6, 611092 (3), Autosomal recessive
GRIN1	100 %	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GRIN2A	100 %	138253	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 (3), Autosomal dominant
GRIN2B	99.99 %	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
GRIN2D	99.97 %	602717	Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant
GRIP1	99.83 %	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GRK1	99.99 %	180381	Oguchi disease-2, 613411 (3)
GRM1	100 %	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
GRM6	100 %	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive
GRM7	99.99 %	604101	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive
GRN	100 %	138945	Frontotemporal dementia 2, 607485 (3), Autosomal dominant, Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant, Autosomal recessive; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
GRXCR1	99.99 %	613283	Deafness, autosomal recessive 25, 613285 (3), Autosomal recessive
GRXCR2	100 %	615762	?Deafness, autosomal recessive 101, 615837 (3), Autosomal recessive
GSC	100 %	138890	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GSDME	99.99 %	608798	Deafness, autosomal dominant 5, 600994 (3), Autosomal dominant
GSN	99.93 %	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
GSR	99.95 %	138300	Hemolytic anemia due to glutathione reductase deficiency, 618660 (3), Autosomal recessive
GSS	99.99 %	601002	Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive
GSTZ1	99.96 %	603758	[Maleylacetoacetate isomerase deficiency], 617596 (3), Autosomal recessive
GSX2	100 %	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
GTF2E2	100 %	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
GTF2H5	100 %	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
GTPBP1	99.9 %	602245	Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888 (3), Autosomal recessive
GTPBP2	99.98 %	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
GTPBP3	99.99 %	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
GUCA1A	100 %	600364	Cone-rod dystrophy 14, 602093 (3), Autosomal dominant; Cone dystrophy-3, 602093 (3), Autosomal dominant
GUCA1B	99.99 %	602275	Retinitis pigmentosa 48, 613827 (3), Autosomal dominant
GUCY1A1	99.99 %	139396	Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive
GUCY2C	99.9 %	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
GUCY2D	100 %	600179	Cone-rod dystrophy 6, 601777 (3), Autosomal dominant, Autosomal recessive; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive
GUF1	99.7 %	617064	?Developmental and epileptic encephalopathy 40, 617065 (3), Autosomal recessive
GUSB	95.07 %	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
GYG1	99.85 %	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
GYPA	98.76 %	617922	{Malaria, resistance to}, 611162 (3); [Blood group, MNSs system], 111300 (3)
GYPB	100 %	617923	[Blood group, Ss], 111740 (3); {Malaria, resistance to}, 611162 (3)
GYPC	100 %	110750	[Blood group, Gerbich], 616089 (3); {Malaria, resistance to}, 611162 (3)
GYS1	99.98 %	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
GYS2	99.86 %	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
GZF1	100 %	613842	Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive
H1-4	100 %	142220	Rahman syndrome, 617537 (3), Autosomal dominant
H3-3A	35.67 %	601128	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 (3), Autosomal dominant
H3-3B	100 %	601058	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 (3), Autosomal dominant
H4C11	99.08 %	602826	?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759 (3), Autosomal dominant
H4C3	99.99 %	602827	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758 (3), Autosomal dominant
H4C5	100 %	602830	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950 (3), Autosomal dominant
H4C9	100 %	602833	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951 (3), Autosomal dominant
H6PD	99.99 %	138090	Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HAAO	99.96 %	604521	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive
HABP2	100 %	603924	{?Thyroid cancer, nonmedullary, 5}, 616535 (3), Autosomal dominant; {Venous thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant
HACD1	99.98 %	610467	Congenital myopathy 11, 619967 (3), Autosomal recessive
HACE1	99.76 %	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
HADH	99.86 %	601609	Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive
HADHA	99.98 %	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency 1, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
HADHB	99.82 %	143450	Mitochondrial trifunctional protein deficiency 2, 620300 (3)
HAGH	100 %	138760	[Glyoxalase II deficiency], 614033 (1), Autosomal dominant
HAL	99.86 %	609457	[Histidinemia], 235800 (3), Autosomal dominant, Autosomal recessive
HAMP	99.99 %	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
HARS1	99.96 %	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
HARS2	99.97 %	600783	Perrault syndrome 2, 614926 (3), Autosomal recessive
HAVCR2	99.93 %	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive
HAX1	100 %	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HBA1	99.95 %	141800	Hemoglobin H disease, nondeletional, 613978 (3); Thalassemias, alpha-, 604131 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant; Methemoglobinemia, alpha type, 617973 (3), Autosomal dominant; Erythrocytosis, familial, 7, 617981 (3), Autosomal dominant
HBA2	72.76 %	141850	Heinz body anemia, 140700 (3), Autosomal dominant; Thalassemia, alpha-, 604131 (3); Erythrocytosis, familial, 7, 617981 (3), Autosomal dominant; Hemoglobin H disease, deletional and nondeletional, 613978 (3)
HBB	100 %	141900	Methemoglobinemia, beta type, 617971 (3), Autosomal dominant; Thalassemia-beta, dominant inclusion-body, 603902 (3), Autosomal dominant; Sickle cell disease, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Delta-beta thalassemia, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; Erythrocytosis, familial, 6, 617980 (3), Autosomal dominant; Heinz body anemia, 140700 (3), Autosomal dominant
HBD	100 %	142000	Thalassemia due to Hb Lepore (3); Thalassemia, delta- (3)
HBEGF	100 %	126150	{Diphtheria, susceptibility to} (1)
HBG1	53.75 %	142200	Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant
HBG2	88.68 %	142250	Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant; Cyanosis, transient neonatal, 613977 (3), Autosomal dominant
HCCS	99.9 %	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HCFC1	99.99 %	300019	Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive
HCK	100 %	142370	Autoinflammation with pulmonary and cutaneous vasculitis, 620296 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HCN1	99.99 %	602780	Developmental and epileptic encephalopathy 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant
HCN2	97.35 %	602781	Febrile seizures, familial, 2, 602477 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 11, 602477 (3), Autosomal dominant
HCN4	100 %	605206	Sick sinus syndrome 2, 163800 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521 (3), Autosomal dominant; Brugada syndrome 8, 613123 (3)
HCRT	99.99 %	602358	?Narcolepsy 1, 161400 (3), Autosomal dominant
HDAC4	99.98 %	605314	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 (3), Autosomal dominant
HDAC6	99.96 %	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
HDAC8	99.74 %	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HDAC9	99.9 %	606543	?Auriculocondylar syndrome 4, 620457 (3), Autosomal dominant
HDC	99.99 %	142704	{Gilles de la Tourette syndrome, susceptibility to}, 137580 (3), Autosomal dominant
HEATR3	99.72 %	614951	Diamond-Blackfan anemia 21, 620072 (3), Autosomal recessive
HECTD4	99.96 %	620209	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive
HECW2	99.91 %	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
HELLS	99.78 %	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive
HEPACAM	100 %	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 (3), Autosomal dominant
HEPHL1	99.99 %	618455	?Abnormal hair, joint laxity, and developmental delay, 261990 (3), Autosomal recessive
HERC1	99.92 %	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive
HERC2	95.66 %	605837	Intellectual developmental disorder, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
HES7	100 %	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
HESX1	99.77 %	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal dominant, Autosomal recessive; Septooptic dysplasia, 182230 (3), Autosomal dominant, Autosomal recessive; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal dominant, Autosomal recessive
HEXA	99.99 %	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HFE	100 %	613609	Hemochromatosis, type 1, 235200 (3), Autosomal recessive
HFM1	89.74 %	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
HGD	99.85 %	607474	Alkaptonuria, 203500 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HGF	99.62 %	142409	Deafness, autosomal recessive 39, 608265 (3), Autosomal recessive
HGSNAT	99.93 %	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HHAT	99.93 %	605743	Nivelon-Nivelon-Mabille syndrome, 600092 (3), Autosomal recessive
HIBCH	99.7 %	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HID1	99.99 %	605752	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 (3), Autosomal recessive
HIKESHI	99.78 %	614908	Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive
HINT1	99.86 %	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive
HIVEP2	99.99 %	143054	Intellectual developmental disorder, autosomal dominant 43, 616977 (3), Autosomal dominant
HJV	99.99 %	608374	Hemochromatosis, type 2A, 602390 (3), Autosomal recessive
HK1	99.97 %	142600	Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive
HKDC1	99.99 %	617221	Retinitis pigmentosa 92, 619614 (3), Autosomal recessive
HLA-A	78.36 %	142800	{Hypersensitivity syndrome, carbamazepine-induced, susceptibility to}, 608579 (3)
HLA-B	75.05 %	142830	{Synovitis, chronic, susceptibility to} (3); {Abacavir hypersensitivity, susceptibility to} (3); {Spondyloarthropathy, susceptibility to, 1}, 106300 (3), Multifactorial; {Stevens-Johnson syndrome, susceptibility to}, 608579 (3); {Drug-induced liver injury due to flucloxacillin} (3); {Toxic epidermal necrolysis, susceptibility to}, 608579 (3)
HLA-C	67.69 %	142840	{Psoriasis susceptibility 1}, 177900 (3), Multifactorial; {HIV-1 viremia, susceptibility to}, 609423 (3)
HLA-DPB1	100 %	142858	{Beryllium disease, chronic, susceptibility to} (3)
HLA-DQA1	84.4 %	146880	{Celiac disease, susceptibility to}, 212750 (3), Multifactorial, Autosomal recessive
HLA-DQB1	97.15 %	604305	{Celiac disease, susceptibility to}, 212750 (3), Multifactorial, Autosomal recessive; {Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Creutzfeldt-Jakob disease, variant, resistance to}, 123400 (3), Autosomal dominant
HLA-DRB1	95.81 %	142857	{Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Sarcoidosis, susceptibility to, 1}, 181000 (3), Autosomal dominant
HLA-G	100 %	142871	{Asthma, susceptibility to}, 600807 (2), Autosomal dominant
HLCS	99.97 %	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMBS	99.97 %	609806	Leukoencephalopathy, porphyria-related, 620711 (3), Autosomal recessive; Encephalopathy, porphyria-related, 620704 (3), Autosomal recessive; Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, 176000 (3), Autosomal dominant
HMCN1	99.44 %	608548	{Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant
HMGA1	100 %	600701	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant
HMGA2	77.65 %	600698	Silver-Russell syndrome 5, 618908 (3), Autosomal dominant
HMGB3	0 %	300193	?Microphthalmia, syndromic 13, 300915 (3), X-linked
HMGCL	99.31 %	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HMGCR	99.73 %	142910	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375 (3), Autosomal recessive; [Statins, response to], 620410 (3); [Low density lipoprotein cholesterol level QTL 3], 620410 (3)
HMGCS2	98.37 %	600234	HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HMMR	99.95 %	600936	{Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant
HMOX1	99.95 %	141250	Heme oxygenase-1 deficiency, 614034 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3)
HMX1	100 %	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
HNF1A	100 %	142410	Hepatic adenoma, somatic, 142330 (3); Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; MODY, type III, 600496 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; Renal cell carcinoma, 144700 (3)
HNF1B	100 %	189907	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
HNF4A	100 %	600281	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant
HNMT	99.53 %	605238	Intellectual developmental disorder, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
HNRNPA1	62.92 %	164017	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 (3), Autosomal dominant; ?Myopathy, distal, 3, 610099 (3), Autosomal dominant; Amyotrophic lateral sclerosis 20, 615426 (3), Autosomal dominant
HNRNPA2B1	99.9 %	600124	Oculopharyngeal muscular dystrophy 2, 620460 (3), Autosomal dominant; ?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3), Autosomal dominant
HNRNPC	100 %	164020	Intellectual developmental disorder, autosomal dominant 74, 620688 (3), Autosomal dominant
HNRNPDL	99.98 %	607137	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115 (3), Autosomal dominant
HNRNPH1	99.98 %	601035	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083 (3), Autosomal dominant
HNRNPH2	99.96 %	300610	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant
HNRNPK	99.93 %	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HNRNPR	98.05 %	607201	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073 (3), Autosomal dominant
HNRNPU	99.95 %	602869	Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant
HOGA1	100 %	613597	Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive
HOMER2	99.99 %	604799	?Deafness, autosomal dominant 68, 616707 (3), Autosomal dominant
HOXA1	100 %	142955	Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive
HOXA11	99.99 %	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
HOXA13	99.94 %	142959	Hand-foot-genital syndrome, 140000 (3), Autosomal dominant; ?Guttmacher syndrome, 176305 (3), Autosomal dominant
HOXA2	100 %	604685	Microtia with or without hearing impairment (AD), 612290 (3), Autosomal dominant, Autosomal recessive; ?Microtia, hearing impairment, and cleft palate (AR), 612290 (3), Autosomal dominant, Autosomal recessive
HOXB1	100 %	142968	Facial paresis, hereditary congenital, 3, 614744 (3), Autosomal recessive
HOXB13	100 %	604607	{Prostate cancer, hereditary, 9}, 610997 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HOXC13	99.98 %	142976	Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive
HOXD10	99.99 %	142984	Vertical talus, congenital, 192950 (3), Autosomal dominant; Charcot-Marie-Tooth disease, foot deformity of, 192950 (3), Autosomal dominant
HOXD13	99.98 %	142989	Syndactyly, type V, 186300 (3), Autosomal dominant; Synpolydactyly 1, 186000 (3), Autosomal dominant; Brachydactyly, type E, 113300 (3), Autosomal dominant; Brachydactyly, type D, 113200 (3), Autosomal dominant; ?Brachydactyly-syndactyly syndrome, 610713 (3)
HP	81.94 %	140100	[Anhaptoglobinemia], 614081 (3); [Hypohaptoglobinemia], 614081 (3)
HPCA	99.98 %	142622	Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive
HPD	99.99 %	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPDL	99.99 %	618994	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive
HPGD	99.98 %	601688	?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive
HPRT1	97.8 %	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HPS1	100 %	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	99.91 %	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	99.98 %	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	99.91 %	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	100 %	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HPSE2	100 %	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
HR	99.95 %	602302	Atrichia with papular lesions, 209500 (3), Autosomal recessive; Alopecia universalis, 203655 (3), Autosomal recessive
HRAS	100 %	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
HRG	99.98 %	142640	Thrombophilia 11 due to HRG deficiency, 613116 (3), Autosomal dominant
HROB	99.98 %	618611	Ovarian dysgenesis 11, 620897 (3), Autosomal recessive
HRURF	100 %	619257	Hypotrichosis 4, 146550 (3), Autosomal dominant
HS2ST1	95.79 %	604844	Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive
HS3ST6	100 %	619210	?Angioedema, hereditary, 8, 619367 (3), Autosomal dominant
HS6ST1	99.99 %	604846	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant
HS6ST2	99.99 %	300545	?Paganini-Miozzo syndrome, 301025 (3), X-linked recessive
HSCB	100 %	608142	?Anemia, sideroblastic, 5, 619523 (3), Autosomal recessive
HSD11B1	99.97 %	600713	Cortisone reductase deficiency 2, 614662 (3), Autosomal dominant
HSD11B2	99.99 %	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
HSD17B10	99.98 %	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HSD17B13	99.68 %	612127	{Fatty liver disease, protection from}, 620116 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HSD17B3	99.77 %	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
HSD17B4	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSD3B2	100 %	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
HSD3B7	100 %	607764	Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive
HSF2BP	99.98 %	604554	Premature ovarian failure 19, 619245 (3), Autosomal recessive
HSF4	99.98 %	602438	Cataract 5, multiple types, 116800 (3), Autosomal dominant
HSPA9	99.96 %	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
HSPB1	99.97 %	602195	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634 (3), Autosomal dominant
HSPB3	99.97 %	604624	?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376 (3), Autosomal dominant
HSPB8	100 %	608014	Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant
HSPD1	83.42 %	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
HSPG2	99.87 %	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
HTR1A	100 %	109760	?Periodic fever, menstrual cycle dependent, 614674 (3), Autosomal dominant
HTR2A	100 %	182135	{Major depressive disorder, response to citalopram therapy in}, 608516 (3); {Obsessive-compulsive disorder, susceptibility to}, 164230 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
HTRA1	100 %	602194	{Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant
HTRA2	99.99 %	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
HTT	99.98 %	613004	Lopes-Maciel-Rodan syndrome, 617435 (3), Autosomal recessive; Huntington disease, 143100 (3), Autosomal dominant
HUWE1	99.88 %	300697	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked
HYAL1	99.99 %	607071	Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive
HYDIN	81.28 %	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
HYLS1	100 %	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
HYOU1	99.97 %	601746	?Immunodeficiency 59 and hypoglycemia, 233600 (3), Autosomal recessive
IARS1	99.89 %	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IARS2	99.77 %	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
IBA57	100 %	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
ICAM1	100 %	147840	{Malaria, cerebral, susceptibility to}, 611162 (3)
ICAM4	100 %	614088	[Blood group, Landsteiner-Wiener], 111250 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ICOS	99.95 %	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
ICOSLG	5.95 %	605717	?Immunodeficiency 119, 620825 (3), Autosomal recessive
IDH1	99.91 %	147700	{Glioma, susceptibility to, somatic}, 137800 (3)
IDH2	100 %	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDH3A	99.98 %	601149	Retinitis pigmentosa 90, 619007 (3), Autosomal recessive
IDH3B	100 %	604526	Retinitis pigmentosa 46, 612572 (3), Autosomal recessive
IDS	99.82 %	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	99.99 %	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
IER3IP1	99.92 %	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFIH1	99.84 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFITM3	100 %	605579	{Influenza, severe, susceptibility to}, 614680 (3)
IFITM5	100 %	614757	Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant
IFNA1	95.03 %	147660	Interferon, alpha, deficiency (1)
IFNAR1	99.75 %	107450	Immunodeficiency 106, susceptibility to viral infections, 619935 (3), Autosomal recessive
IFNAR2	89.62 %	602376	{Hepatitis B virus, susceptibility to}, 610424 (3); Immunodeficiency 45, 616669 (3), Autosomal recessive
IFNG	99.5 %	147570	{Hepatitis C virus, response to therapy of}, 609532 (3); {TSC2 angiomyolipomas, renal, modifier of}, 613254 (3), Autosomal dominant; {Aplastic anemia}, 609135 (3); ?Immunodeficiency 69, mycobacteriosis, 618963 (3), Autosomal recessive; {Tuberculosis, protection against}, 607948 (3); {AIDS, rapid progression to}, 609423 (3)
IFNGR1	99.87 %	107470	{H. pylori infection, susceptibility to}, 600263 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3)
IFNGR2	99.95 %	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
IFNL3	100 %	607402	{Hepatitis C virus infection, response to therapy of}, 609532 (3)
IFT122	99.98 %	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	100 %	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
IFT172	99.98 %	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	100 %	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	99.97 %	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive
IFT52	99.82 %	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
IFT57	99.43 %	606621	?Orofaciodigital syndrome XVIII, 617927 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IFT74	99.71 %	608040	Bardet-Biedl syndrome 22, 617119 (3), Autosomal recessive; Spermatogenic failure 58, 619585 (3), Autosomal recessive; Joubert syndrome 40, 619582 (3), Autosomal recessive
IFT80	99.69 %	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IFT81	94.64 %	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
IGBP1	99.9 %	300139	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
IGF1	100 %	147440	Insulin-like growth factor I deficiency, 608747 (3), Autosomal recessive
IGF1R	100 %	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal dominant, Autosomal recessive
IGF2	100 %	147470	Silver-Russell syndrome 3, 616489 (3), Autosomal dominant
IGF2BP2	100 %	608289	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
IGF2R	99.99 %	147280	Hepatocellular carcinoma, somatic, 114550 (3)
IGFALS	100 %	601489	Acid-labile subunit, deficiency of, 615961 (3), Autosomal recessive
IGFBP7	99.97 %	602867	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3), Autosomal recessive
IGHG2	99.93 %	147110	IgG2 deficiency, selective (3)
IGHM	100 %	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
IGHMBP2	99.92 %	600502	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320 (3), Autosomal recessive
IGKC	99.99 %	147200	Kappa light chain deficiency, 614102 (3), Autosomal recessive
IGLL1	100 %	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
IGSF1	99.98 %	300137	Hypothyroidism, central, and testicular enlargement, 300888 (3), X-linked recessive
IGSF3	99.94 %	603491	?Lacrimal duct defect, 149700 (3), Autosomal recessive
IHH	100 %	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
IKBKB	99.93 %	603258	Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant
IKBKG	57.34 %	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
IKZF1	99.92 %	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IKZF3	99.96 %	606221	?Immunodeficiency 84, 619437 (3), Autosomal dominant
IKZF5	99.6 %	606238	Thrombocytopenia, autosomal dominant, 7, 619130 (3), Autosomal dominant
IL10	100 %	124092	{Rheumatoid arthritis, progression of}, 180300 (3); {Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3)
IL10RA	99.99 %	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
IL10RB	99.99 %	123889	{Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive
IL11RA	99.98 %	600939	Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive
IL12B	99.98 %	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IL12RB1	94.11 %	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
IL13	100 %	147683	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Allergic rhinitis, susceptibility to}, 607154 (3)
IL17F	99.99 %	606496	?Candidiasis, familial, 6, autosomal dominant, 613956 (3)
IL17RA	100 %	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
IL17RC	100 %	610925	Candidiasis, familial, 9, 616445 (3), Autosomal recessive
IL17RD	99.94 %	606807	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
IL18BP	99.99 %	604113	{?Hepatitis, fulminant viral, susceptibility to}, 618549 (3), Autosomal recessive
IL1B	99.96 %	147720	{Gastric cancer risk after H. pylori infection}, 613659 (3)
IL1R1	97.25 %	147810	?Chronic recurrent multifocal osteomyelitis 3, 259680 (3), Autosomal dominant
IL1RAPL1	99.77 %	300206	Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive
IL1RN	99.64 %	147679	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852 (3), Autosomal recessive; {Gastric cancer risk after H. pylori infection}, 613659 (3); {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
IL21	99.95 %	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
IL21R	99.7 %	605383	Immunodeficiency 56, 615207 (3), Autosomal recessive
IL23R	97.64 %	607562	{Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3)
IL2RA	99.99 %	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
IL2RB	100 %	146710	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive
IL2RG	99.86 %	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive
IL31RA	100 %	609510	?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant
IL36RN	100 %	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
IL37	100 %	605510	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398 (3), Autosomal recessive
IL6	100 %	147620	{Type 2 diabetes mellitus}, 125853 (3), Autosomal dominant; {Rheumatoid arthritis, systemic juvenile}, 604302 (3); {Intracranial hemorrhage in brain cerebrovascular malformations, susceptibility to}, 108010 (3), Somatic mutation; {Type 1 diabetes mellitus}, 222100 (3), Autosomal recessive; {Kaposi sarcoma in HIV+, susceptibility to}, 148000 (3), Autosomal dominant; {Crohn disease-associated growth failure}, 266600 (3), Multifactorial
IL6R	92.46 %	147880	[Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE syndrome 5, autosomal recessive, with recurrent infections, 618944 (3), Autosomal recessive; [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3)
IL6ST	99.88 %	600694	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752 (3), Autosomal dominant; Stuve-Wiedemann syndrome 2, 619751 (3), Autosomal recessive; Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523 (3), Autosomal recessive; ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 (3), Autosomal dominant
IL7	99.57 %	146660	{?Epidermodysplasia verruciformis, susceptibility to, 5}, 618309 (3), Autosomal recessive
IL7R	99.99 %	146661	Immunodeficiency 104, severe combined, 608971 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ILDR1	99.97 %	609739	Deafness, autosomal recessive 42, 609646 (3), Autosomal recessive
IMPA1	99.95 %	602064	Intellectual developmental disorder, autosomal recessive 59, 617323 (3), Autosomal recessive
IMPDH1	92.77 %	146690	Retinitis pigmentosa 10, 180105 (3), Autosomal dominant; Leber congenital amaurosis 11, 613837 (3), Autosomal dominant
IMPDH2	100 %	146691	[IMPDH2 enzyme activity, variation in], 617995 (3)
IMPG1	99.93 %	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant, Autosomal recessive; Retinitis pigmentosa 91, 153870 (3), Autosomal dominant
IMPG2	99.97 %	607056	Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant
INAVA	99.93 %	618051	{Inflammatory bowel disease 29}, 618077 (3), Autosomal dominant
INF2	99.99 %	610982	Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant
ING1	100 %	601566	Squamous cell carcinoma, head and neck, somatic, 275355 (3)
INPP5E	99.85 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INPP5K	99.94 %	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
INPPL1	99.97 %	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
INS	100 %	176730	Diabetes mellitus, insulin-dependent, 2, 125852 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 10, 613370 (3), Autosomal dominant; Hyperproinsulinemia, 616214 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 4, 618858 (3), Autosomal dominant, Autosomal recessive
INSL3	98.02 %	146738	Cryptorchidism, 219050 (3), Autosomal dominant
INSR	99.99 %	147670	Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive; Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Donohue syndrome, 246200 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant
INTS1	100 %	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive
INTS11	100 %	611354	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 (3), Autosomal recessive
INTS8	99.89 %	611351	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive
INTU	99.92 %	610621	?Orofaciodigital syndrome XVII, 617926 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 20 with polydactyly, 617925 (3), Autosomal recessive
INVS	99.94 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IPO8	97.96 %	605600	VISS syndrome, 619472 (3), Autosomal recessive
IQCB1	99.72 %	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
IQCE	99.77 %	617631	Polydactyly, postaxial, type A7, 617642 (3), Autosomal recessive
IQCN	100 %	620160	Spermatogenic failure 78, 620170 (3), Autosomal recessive
IQSEC1	99.99 %	610166	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive
IQSEC2	99.97 %	300522	Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant
IRAK3	99.36 %	604459	{Asthma susceptibility 5}, 611064 (3)
IRAK4	98.85 %	606883	Immunodeficiency 67, 607676 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IREB2	99.9 %	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
IRF1	99.98 %	147575	Nonsmall cell lung cancer, somatic, 211980 (3); Gastric cancer, somatic, 613659 (3); Immunodeficiency 117, mycobacteriosis, autosomal recessive, 620668 (3), Autosomal recessive
IRF2BP2	100 %	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant
IRF2BPL	99.21 %	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
IRF3	99.96 %	603734	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 (3), Autosomal dominant
IRF4	99.99 %	601900	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
IRF5	99.95 %	607218	{Inflammatory bowel disease 14}, 612245 (3); {Systemic lupus erythematosus, susceptibility to, 10}, 612251 (3)
IRF6	99.96 %	607199	{Orofacial cleft 6}, 608864 (3), Autosomal dominant; Popliteal pterygium syndrome 1, 119500 (3), Autosomal dominant; van der Woude syndrome 1, 119300 (3), Autosomal dominant
IRF7	100 %	605047	?Immunodeficiency 39, 616345 (3), Autosomal recessive
IRF8	99.99 %	601565	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive
IRF9	100 %	147574	Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive
IRGM	100 %	608212	{Mycobacterium tuberculosis, protection against}, 607948 (3); {Inflammatory bowel disease (Crohn disease) 19}, 612278 (3)
IRS1	100 %	147545	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; {Coronary artery disease, susceptibility to} (3)
IRS2	100 %	600797	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
IRS4	99.98 %	300904	Hypothyroidism, congenital, nongoitrous, 9, 301035 (3), X-linked recessive
IRX5	100 %	606195	Hamamy syndrome, 611174 (3), Autosomal recessive
ISCA1	99.79 %	611006	Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive
ISCA2	100 %	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
ISCU	99.74 %	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive
ISG15	100 %	147571	Immunodeficiency 38, 616126 (3), Autosomal recessive
ITCH	95.57 %	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
ITGA2B	99.99 %	607759	Thrombocytopenia, neonatal alloimmune, BAK antigen related (3); Glanzmann thrombasthenia 1, 273800 (3), Autosomal recessive; Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant
ITGA3	99.86 %	605025	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 (3), Autosomal recessive
ITGA6	99.78 %	147556	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817 (3), Autosomal recessive
ITGA7	99.87 %	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
ITGA8	99.95 %	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive
ITGB2	100 %	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ITGB3	99.83 %	173470	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 (3), Autosomal dominant; Thrombocytopenia, neonatal alloimmune (3); Purpura, posttransfusion (3); {Myocardial infarction, susceptibility to}, 608446 (3); Glanzmann thrombasthenia 2, 619267 (3), Autosomal recessive
ITGB4	99.99 %	147557	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive
ITGB6	99.71 %	147558	Amelogenesis imperfecta, type IH, 616221 (3), Autosomal recessive
ITK	99.91 %	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
ITM2B	99.85 %	603904	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
ITPA	99.99 %	147520	[Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive
ITPR1	99.98 %	147265	Gillespie syndrome, 206700 (3), Autosomal dominant, Autosomal recessive; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant
ITPR2	99.13 %	600144	?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive
ITPR3	99.98 %	147267	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111 (3), Autosomal dominant; {Diabetes, type 1, susceptibility to}, 222100 (2), Autosomal recessive
IVD	100 %	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
IVNS1ABP	99.24 %	609209	Immunodeficiency 70, 618969 (3), Autosomal dominant
IYD	100 %	612025	Thyroid dyshormonogenesis 4, 274800 (3), Autosomal recessive
JAG1	100 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAG2	99.99 %	602570	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566 (3), Autosomal recessive
JAGN1	100 %	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
JAK1	99.32 %	147795	Autoinflammation, immune dysregulation, and eosinophilia, 618999 (3), Autosomal dominant
JAK2	99.52 %	147796	{Budd-Chiari syndrome, somatic}, 600880 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3); Leukemia, acute myeloid, somatic, 601626 (3); Thrombocythemia 3, 614521 (3), Somatic mutation, Autosomal dominant; Polycythemia vera, somatic, 263300 (3)
JAK3	99.99 %	600173	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
JAM2	91.82 %	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
JAM3	100 %	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JARID2	99.97 %	601594	Developmental delay with variable intellectual disability and dysmorphic facies, 620098 (3), Autosomal dominant
JPH1	99.98 %	605266	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, modifier of, 607831 (3), Autosomal dominant, Autosomal recessive
JPH2	99.99 %	605267	Cardiomyopathy, dilated, 2E, 619492 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 17, 613873 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
JPH3	100 %	605268	Huntington disease-like 2, 606438 (3), Autosomal dominant
JUP	99.94 %	173325	Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant
KANK1	99.99 %	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANK2	99.99 %	614610	Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive; Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive
KANSL1	99.85 %	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
KARS1	99.98 %	601421	Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive
KASH5	99.97 %	618125	Spermatogenic failure 88, 620547 (3), Autosomal recessive; Premature ovarian failure 22, 620548 (3)
KAT5	100 %	601409	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3), Autosomal dominant
KAT6A	99.93 %	601408	Arboleda-Tham syndrome, 616268 (3), Autosomal dominant
KAT6B	99.79 %	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
KAT8	99.98 %	609912	Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant
KATNB1	99.99 %	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
KATNIP	99.13 %	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
KBTBD13	100 %	613727	Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant
KCNA1	100 %	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA2	99.99 %	176262	Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant
KCNA4	100 %	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCNA5	100 %	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
KCNB1	100 %	600397	Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant
KCNC1	100 %	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
KCNC2	99.88 %	176256	Developmental and epileptic encephalopathy 103, 619913 (3), Autosomal dominant
KCNC3	99.98 %	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
KCND3	99.98 %	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant
KCNE1	87.07 %	176261	Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive; Long QT syndrome 5, 613695 (3), Autosomal dominant
KCNE2	99.99 %	603796	Long QT syndrome 6, 613693 (3), Autosomal dominant; Atrial fibrillation, familial, 4, 611493 (3)
KCNE3	100 %	604433	?Brugada syndrome 6, 613119 (3)
KCNH1	99.92 %	603305	Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant
KCNH2	99.99 %	152427	Short QT syndrome 1, 609620 (3); Long QT syndrome 2, 613688 (3), Autosomal dominant
KCNH5	99.89 %	605716	Developmental and epileptic encephalopathy 112, 620537 (3), Autosomal dominant
KCNJ1	100 %	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
KCNJ10	99.98 %	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KCNJ11	100 %	600937	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal dominant, Autosomal recessive
KCNJ13	99.99 %	603208	Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive
KCNJ16	100 %	605722	Hypokalemic tubulopathy and deafness, 619406 (3), Autosomal recessive
KCNJ18	100 %	613236	{Thyrotoxic periodic paralysis, susceptibility to, 2}, 613239 (3), Autosomal dominant
KCNJ2	100 %	600681	Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3), Autosomal dominant
KCNJ5	99.99 %	600734	Long QT syndrome 13, 613485 (3), Autosomal dominant; Hyperaldosteronism, familial, type III, 613677 (3), Autosomal dominant
KCNJ6	100 %	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNK18	100 %	613655	{Migraine, with or without aura, susceptibility to, 13}, 613656 (3), Autosomal dominant
KCNK3	100 %	603220	Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant
KCNK4	99.99 %	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant
KCNK9	100 %	605874	Birk-Barel syndrome, 612292 (3)
KCNMA1	99.89 %	600150	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant
KCNMB1	99.98 %	603951	{Hypertension, diastolic, resistance to}, 608622 (3), Autosomal dominant
KCNN2	91.25 %	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
KCNN3	99.97 %	602983	Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant
KCNN4	99.98 %	602754	Dehydrated hereditary stomatocytosis 2, 616689 (3), Autosomal dominant
KCNQ1	100 %	607542	Short QT syndrome 2, 609621 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive
KCNQ1OT1	0.38 %	604115	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
KCNQ2	100 %	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCNQ3	99.98 %	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
KCNQ4	99.19 %	603537	Deafness, autosomal dominant 2A, 600101 (3), Autosomal dominant
KCNQ5	99.89 %	607357	Intellectual developmental disorder, autosomal dominant 46, 617601 (3), Autosomal dominant
KCNT1	99.98 %	608167	Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant
KCNT2	98.77 %	610044	Developmental and epileptic encephalopathy 57, 617771 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KCNU1	99.93 %	615215	Spermatogenic failure 79, 620196 (3), Autosomal recessive
KCNV2	99.97 %	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
KCTD1	99.99 %	613420	Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant
KCTD17	100 %	616386	Dystonia 26, myoclonic, 616398 (3), Autosomal dominant
KCTD7	99.98 %	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KDELR2	99.98 %	609024	Osteogenesis imperfecta, type XXI, 619131 (3), Autosomal recessive
KDF1	99.86 %	616758	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant
KDM1A	89.42 %	609132	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
KDM3B	99.98 %	609373	Diets-Jongmans syndrome, 618846 (3), Autosomal dominant
KDM4B	99.99 %	609765	Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant
KDM5A	99.9 %	180202	El Hayek-Chahrour neurodevelopmental syndrome, 620820 (3), Autosomal recessive
KDM5B	97.15 %	605393	Intellectual developmental disorder, autosomal recessive 65, 618109 (3), Autosomal recessive
KDM5C	99.98 %	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM6A	99.74 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KDM6B	99.99 %	611577	Stolerman neurodevelopmental syndrome, 618505 (3), Autosomal dominant
KDR	99.86 %	191306	{Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3)
KDSR	99.99 %	136440	Erythrokeratodermia variabilis et progressiva 4, 617526 (3), Autosomal recessive
KEL	99.98 %	613883	[Blood group, Kell], 110900 (3)
KERA	99.95 %	603288	Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive
KHDC3L	100 %	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
KHK	99.99 %	614058	?[Fructosuria, essential], 229800 (3), Autosomal recessive
KIAA0586	95.75 %	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
KIAA0753	100 %	617112	?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive; ?Joubert syndrome 38, 619476 (3), Autosomal recessive; Short-rib thoracic dysplasia 21 without polydactyly, 619479 (3), Autosomal recessive
KIAA0825	99.9 %	617266	Polydactyly, postaxial, type A10, 618498 (3), Autosomal recessive
KIAA1109	99.79 %	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
KIAA1549	99.98 %	613344	Retinitis pigmentosa 86, 618613 (3), Autosomal recessive
KIDINS220	99.94 %	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogryposis, 619501 (3), Autosomal recessive
KIF11	99.84 %	148760	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 (3), Autosomal dominant
KIF12	99.99 %	611278	Cholestasis, progressive familial intrahepatic, 8, 619662 (3), Autosomal recessive
KIF14	97.8 %	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
KIF15	99.93 %	617569	?Braddock-Carey syndrome 2, 619981 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KIF1A	99.96 %	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant; Spastic paraplegia 30, autosomal recessive, 620607 (3), Autosomal recessive
KIF1B	99.97 %	605995	{Neuroblastoma, susceptibility to, 1}, 256700 (3), Somatic mutation, Autosomal dominant; Charcot-Marie-Tooth disease, type 2A1, 118210 (3), Autosomal dominant
KIF1C	99.99 %	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
KIF20A	100 %	605664	?Cardiomyopathy, familial restrictive, 6, 619433 (3), Autosomal recessive
KIF21A	96.41 %	608283	Fibrosis of extraocular muscles, congenital, 3B, 135700 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 1, 135700 (3), Autosomal dominant
KIF22	99.89 %	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant
KIF23	99.82 %	605064	Anemia, congenital dyserythropoietic, type IIIA, 105600 (3), Autosomal dominant
KIF26A	100 %	613231	Cortical dysplasia, complex, with other brain malformations 11, 620156 (3), Autosomal recessive
KIF2A	99.79 %	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
KIF3B	99.99 %	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
KIF4A	99.76 %	300521	Taurodontism, microdontia, and dens invaginatus, 313490 (3), X-linked recessive; Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive
KIF5A	99.91 %	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
KIF5C	99.93 %	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
KIF7	100 %	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KIFBP	99.91 %	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
KIRREL1	99.82 %	607428	Nephrotic syndrome, type 23, 619201 (3), Autosomal recessive
KISS1	99.97 %	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive
KISS1R	100 %	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
KIT	99.86 %	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Isolated cases, Autosomal dominant; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Piebaldism, 172800 (3), Autosomal dominant; Germ cell tumors, somatic, 273300 (3); Mastocytosis, systemic, somatic, 154800 (3); Leukemia, acute myeloid, somatic, 601626 (3)
KITLG	99.46 %	184745	Hyperpigmentation with or without hypopigmentation, 145250 (3), Autosomal dominant; Waardenburg syndrome, type 2F, 619947 (3), Autosomal recessive; Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 (3), Autosomal dominant; [Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3)
KIZ	99.98 %	615757	Retinitis pigmentosa 69, 615780 (3), Autosomal recessive
KL	99.98 %	604824	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3), Autosomal recessive
KLC2	100 %	611729	Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive
KLF1	100 %	600599	Blood group--Lutheran inhibitor, 111150 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type IV, 613673 (3), Autosomal dominant; [Hereditary persistence of fetal hemoglobin], 613566 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KLF11	99.99 %	603301	Maturity-onset diabetes of the young, type VII, 610508 (3)
KLF6	100 %	602053	Gastric cancer, somatic, 613659 (3); Prostate cancer, somatic, 176807 (3)
KLHDC8B	99.97 %	613169	{Hodgkin lymphoma, susceptibility to}, 236000 (3), Autosomal recessive
KLHL10	99.97 %	608778	Spermatogenic failure 11, 615081 (3), Autosomal dominant
KLHL15	100 %	300980	Intellectual developmental disorder, X-linked 103, 300982 (3), X-linked recessive
KLHL24	99.9 %	611295	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236 (3), Autosomal recessive; Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294 (3), Autosomal dominant
KLHL3	99.97 %	605775	Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal dominant, Autosomal recessive
KLHL40	99.96 %	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive
KLHL41	99.96 %	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
KLHL7	99.95 %	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
KLK1	100 %	147910	[Kallikrein, decreased urinary activity of], 615953 (3)
KLK11	99.96 %	604434	Ichthyosis with erythrokeratoderma, 620507 (3), Autosomal dominant
KLK4	99.99 %	603767	Amelogenesis imperfecta, type IIA1, 204700 (3), Autosomal recessive
KLKB1	99.96 %	229000	Fletcher factor (prekallikrein) deficiency, 612423 (3), Autosomal recessive
KLLN	100 %	612105	Cowden syndrome 4, 615107 (3)
KMT2A	99.97 %	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2B	99.99 %	606834	Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
KMT2C	98.98 %	606833	Kleefstra syndrome 2, 617768 (3), Autosomal dominant
KMT2D	99.98 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant
KMT2E	99.92 %	608444	O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant
KMT5B	99.91 %	610881	Intellectual developmental disorder, autosomal dominant 51, 617788 (3), Autosomal dominant
KNG1	99.99 %	612358	[Kininogen deficiency], 228960 (3), Autosomal recessive; Angioedema, hereditary, 6, 619363 (3), Autosomal dominant; [High molecular weight kininogen deficiency], 228960 (3), Autosomal recessive
KNL1	98.43 %	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
KNSTRN	100 %	614718	?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive
KPNA3	99.89 %	601892	Spastic paraplegia 88, autosomal dominant, 620106 (3), Autosomal dominant
KPNA7	99.51 %	614107	Oocyte/zygote/embryo maturation arrest 17, 620319 (3), Autosomal recessive
KPTN	99.98 %	615620	Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive
KRAS	99.13 %	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)
KREMEN1	99.88 %	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KRIT1	99.33 %	604214	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Cavernous malformations of CNS and retina, 116860 (3), Autosomal dominant
KRT1	99.89 %	139350	Ichthyosis, annular epidermolytic 2, 620148 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, 600962 (3), Autosomal dominant; Epidermolytic hyperkeratosis 1, 113800 (3), Autosomal dominant; Palmoplantar keratoderma, epidermolytic, 2, 620411 (3), Autosomal dominant; Keratosis palmoplantaris striata III, 607654 (3); Ichthyosis histrix, Curth-Macklin type, 146590 (3), Autosomal dominant
KRT10	100 %	148080	Ichthyosis, annular epidermolytic 1, 607602 (3), Autosomal dominant; Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707 (3), Autosomal recessive; Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150 (3), Autosomal dominant; ?Ichthyosis histrix, Lambert type, 146600 (3), Autosomal dominant; Ichthyosis with confetti, 609165 (3), Autosomal dominant
KRT12	99.92 %	601687	Meesmann corneal dystrophy 1, 122100 (3), Autosomal dominant
KRT13	99.98 %	148065	White sponge nevus 2, 615785 (3), Autosomal dominant
KRT14	99.99 %	148066	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex 1C, localized, 131800 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex 1A, generalized severe, 131760 (3), Autosomal dominant; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex 1B, generalized intermediate, 131900 (3), Autosomal dominant
KRT16	100 %	148067	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant; Pachyonychia congenita 1, 167200 (3), Autosomal dominant
KRT17	100 %	148069	Steatocystoma multiplex, 184500 (3), Autosomal dominant; Pachyonychia congenita 2, 167210 (3), Autosomal dominant
KRT18	47.42 %	148070	Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive
KRT2	99.88 %	600194	Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant
KRT25	99.77 %	616646	Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive
KRT3	99.89 %	148043	Meesmann corneal dystrophy 2, 618767 (3), Autosomal dominant
KRT4	99.98 %	123940	White sponge nevus 1, 193900 (3), Autosomal dominant
KRT5	99.98 %	148040	Epidermolysis bullosa simplex 2A, generalized severe, 619555 (3), Autosomal dominant; Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 (3), Autosomal dominant; Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 (3), Autosomal recessive; Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 (3), Autosomal dominant; Epidermolysis bullosa simplex 2C, localized, 619594 (3), Autosomal dominant; Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352 (3), Autosomal dominant
KRT6A	100 %	148041	Pachyonychia congenita 3, 615726 (3), Autosomal dominant
KRT6B	99.95 %	148042	Pachyonychia congenita 4, 615728 (3), Autosomal dominant
KRT6C	96.59 %	612315	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant
KRT71	99.96 %	608245	?Hypotrichosis 13, 615896 (3), Autosomal dominant
KRT74	99.92 %	608248	Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; ?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KRT75	99.99 %	609025	{Pseudofolliculitis barbae, susceptibility to}, 612318 (3)
KRT81	78.07 %	602153	Monilethrix, 158000 (3), Autosomal dominant
KRT83	100 %	602765	Monilethrix, 158000 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 5, 617756 (3), Autosomal recessive
KRT85	99.97 %	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
KRT86	77.97 %	601928	Monilethrix, 158000 (3), Autosomal dominant
KRT9	100 %	607606	Palmoplantar keratoderma, epidermolytic, 1, 144200 (3), Autosomal dominant
KY	99.97 %	605739	Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive
KYNU	99.19 %	605197	?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive
L1CAM	99.98 %	308840	MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus, congenital, X-linked, 307000 (3), X-linked recessive; ?Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive
L2HGDH	99.92 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LACC1	99.99 %	613409	Juvenile arthritis, 618795 (3), Autosomal recessive
LAGE3	99.99 %	300060	Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive
LAMA1	99.98 %	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMA2	99.95 %	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
LAMA3	99.9 %	600805	Epidermolysis bullosa, junctional 2A, intermediate, 619783 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2B, severe, 619784 (3), Autosomal recessive
LAMA4	99.93 %	600133	Cardiomyopathy, dilated, 1JJ, 615235 (3), Autosomal dominant
LAMA5	99.99 %	601033	Nephrotic syndrome, type 26, 620049 (3), Autosomal recessive; ?Bent bone dysplasia syndrome 2, 620076 (3), Autosomal recessive
LAMB1	99.87 %	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
LAMB2	99.99 %	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3), Autosomal recessive; Pierson syndrome, 609049 (3), Autosomal recessive
LAMB3	99.99 %	150310	Epidermolysis bullosa, junctional 1B, severe, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional 1A, intermediate, 226650 (3), Autosomal recessive; Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant
LAMC2	99.54 %	150292	Epidermolysis bullosa, junctional 3B, severe, 619786 (3), Autosomal recessive; Epidermolysis bullosa, junctional 3A, intermediate, 619785 (3), Autosomal recessive
LAMC3	99.97 %	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
LAMP2	98.95 %	309060	Danon disease, 300257 (3), X-linked dominant
LAMTOR2	99.92 %	610389	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive
LARGE1	100 %	603590	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
LARP7	99.63 %	612026	Alazami syndrome, 615071 (3), Autosomal recessive
LARS1	99.87 %	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
LARS2	99.96 %	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LAS1L	99.97 %	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LAT	99.85 %	602354	Immunodeficiency 52, 617514 (3), Autosomal recessive
LBR	99.66 %	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 (3), Autosomal recessive; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive
LBX1	99.99 %	604255	?Central hypoventilation syndrome, congenital, 3, 619483 (3), Autosomal recessive
LCA5	99.89 %	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
LCAT	99.97 %	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
LCK	99.56 %	153390	Immunodeficiency 22, 615758 (3), Autosomal recessive
LCP2	99.58 %	601603	Immunodeficiency 81, 619374 (3), Autosomal recessive
LCT	99.98 %	603202	Lactase deficiency, congenital, 223000 (3), Autosomal recessive
LDB3	99.91 %	605906	Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant; Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant
LDHA	99.94 %	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
LDHB	99.84 %	150100	[Lactate dehydrogenase-B deficiency], 614128 (3)
LDHD	99.96 %	607490	D-lactic aciduria with susceptibility to gout, 245450 (3), Autosomal recessive
LDLR	99.99 %	606945	LDL cholesterol level QTL2, 143890 (3), Autosomal dominant, Autosomal recessive; Hypercholesterolemia, familial, 1, 143890 (3), Autosomal dominant, Autosomal recessive
LDLRAP1	99.89 %	605747	Hypercholesterolemia, familial, 4, 603813 (3), Autosomal recessive
LEF1	99.96 %	153245	Sebaceous tumors, somatic (3)
LEMD2	100 %	616312	Marbach-Rustad progeroid syndrome, 619322 (3), Autosomal dominant; Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive
LEMD3	99.07 %	607844	Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant; Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant
LEP	100 %	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
LEPR	92.52 %	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive
LETM1	99.97 %	604407	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive
LFNG	100 %	602576	Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive
LGALS2	100 %	150571	{Myocardial infarction, susceptibility to}, 608446 (3)
LGI1	99.99 %	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
LGI3	100 %	608302	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007 (3), Autosomal recessive
LGI4	99.98 %	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
LGR4	99.96 %	606666	Delayed puberty, self-limited, 619613 (3), Autosomal dominant; {Bone mineral density, low, susceptibility to}, 615311 (3), Autosomal dominant
LHB	99.93 %	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LHCGR	99.94 %	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant
LHFPL5	100 %	609427	Deafness, autosomal recessive 67, 610265 (3), Autosomal recessive
LHX3	99.99 %	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
LHX4	99.99 %	602146	Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant
LIAS	99.98 %	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
LIFR	99.69 %	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
LIG1	99.93 %	126391	Immunodeficiency 96, 619774 (3), Autosomal recessive
LIG3	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
LIG4	100 %	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LIM2	100 %	154045	Cataract 19, multiple types, 615277 (3), Autosomal dominant, Autosomal recessive
LIMA1	99.8 %	608364	[Low density lipoprotein cholesterol level QTL 8], 618079 (3)
LIMS2	99.97 %	607908	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 (3), Autosomal recessive
LINGO1	100 %	609791	Intellectual developmental disorder, autosomal recessive 64, 618103 (3), Autosomal recessive
LINS1	99.96 %	610350	Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive
LIPA	99.96 %	613497	Wolman disease, 620151 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
LIPC	99.89 %	151670	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Hepatic lipase deficiency, 614025 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 12], 612797 (3)
LIPE	100 %	151750	Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive
LIPH	99.99 %	607365	Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive
LIPN	99.94 %	613924	Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive
LIPT1	99.89 %	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
LIPT2	99.99 %	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
LITAF	83.22 %	603795	Charcot-Marie-Tooth disease, type 1C, 601098 (3), Autosomal dominant
LMAN1	99.96 %	601567	Combined factor V and VIII deficiency, 227300 (3), Autosomal recessive
LMAN2L	99.91 %	609552	?Intellectual developmental disorder, autosomal dominant 69, 617863 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 52, 616887 (3), Autosomal recessive
LMBR1	99.88 %	605522	Syndactyly, type IV, 186200 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Acheiropody, 200500 (3), Autosomal recessive; Triphalangeal thumb-polysyndactyly syndrome, 190605 (3), Autosomal dominant
LMBRD1	99.67 %	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive
LMBRD2	99.66 %	619490	Developmental delay with variable neurologic and brain abnormalities, 619694 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LMF1	100 %	611761	Lipase deficiency, combined, 246650 (3), Autosomal recessive
LMLN2	91.91 %	619703	Heterotaxy, visceral, 12, autosomal, 619702 (3), Autosomal recessive
LMNA	99.96 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
LMNB1	99.73 %	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant
LMNB2	99.99 %	150341	Microcephaly 27, primary, autosomal dominant, 619180 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant
LMO1	99.94 %	186921	Leukemia, T-cell acute lymphoblastic, 186921 (2)
LMO2	100 %	180385	Leukemia, acute T-cell, 180385 (2)
LMOD1	100 %	602715	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362 (3), Autosomal recessive
LMOD2	99.96 %	608006	Cardiomyopathy, dilated, 2G, 619897 (3), Autosomal recessive
LMOD3	99.91 %	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
LMX1A	99.94 %	600298	Deafness, autosomal dominant 7, 601412 (3), Autosomal dominant
LMX1B	100 %	602575	Focal segmental glomerulosclerosis 10, 256020 (3), Autosomal dominant; Nail-patella syndrome, 161200 (3), Autosomal dominant
LNPK	92.81 %	610236	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive
LONP1	99.99 %	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LORICRIN	100 %	152445	Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant
LOX	99.85 %	153455	Aortic aneurysm, familial thoracic 10, 617168 (3), Autosomal dominant
LOXHD1	99.99 %	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
LOXL1	99.81 %	153456	{Exfoliation syndrome, susceptibility to}, 177650 (3), Autosomal dominant
LOXL3	99.95 %	607163	Myopia 28, autosomal recessive, 619781 (3), Autosomal recessive
LPA	69.68 %	152200	[LPA deficiency, congenital], 618807 (3), Autosomal dominant; {Coronary artery disease, susceptibility to}, 618807 (3), Autosomal dominant
LPAR6	99.99 %	609239	Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive
LPIN1	99.96 %	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive
LPIN2	100 %	605519	Majeed syndrome, 609628 (3)
LPL	100 %	609708	Lipoprotein lipase deficiency, 238600 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 11], 238600 (3), Autosomal recessive; Combined hyperlipidemia, familial, 144250 (3), Autosomal dominant
LPP	99.65 %	600700	Lipoma (3); Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LRAT	100 %	604863	Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive
LRBA	99.76 %	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
LRIF1	99.82 %	615354	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477 (3), Digenic recessive
LRIG2	97.97 %	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
LRIT3	100 %	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
LRMDA	99.87 %	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
LRP1	99.94 %	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive; Developmental dysplasia of the hip 3, 620690 (3), Autosomal dominant
LRP12	99.99 %	618299	Oculopharyngodistal myopathy 1, 164310 (3), Autosomal dominant; Amyotrophic lateral sclerosis 28, 620452 (3), Autosomal dominant
LRP2	99.86 %	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP4	99.89 %	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal dominant, Autosomal recessive; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
LRP5	99.95 %	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Exudative vitreoretinopathy 4, 601813 (3), Autosomal dominant, Autosomal recessive
LRP6	99.91 %	603507	{Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant
LRP8	99.24 %	602600	{Myocardial infarction, susceptibility to}, 608446 (3)
LRPAP1	99.99 %	104225	Myopia 23, autosomal recessive, 615431 (3), Autosomal recessive
LRPPRC	99.8 %	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive
LRRC23	100 %	620708	Spermatogenic failure 92, 620848 (3), Autosomal recessive
LRRC32	100 %	137207	Cleft palate, proliferative retinopathy, and developmental delay, 619074 (3), Autosomal recessive
LRRC56	99.98 %	618227	Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive
LRRC8A	100 %	608360	?Agammaglobulinemia 5, 613506 (3), Autosomal dominant
LRRK1	99.99 %	610986	Osteosclerotic metaphyseal dysplasia, 615198 (3), Autosomal recessive
LRRK2	99.44 %	609007	{Parkinson disease 8}, 607060 (3), Autosomal dominant
LRSAM1	99.99 %	610933	Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal dominant, Autosomal recessive
LRTOMT	99.99 %	612414	Deafness, autosomal recessive 63, 611451 (3), Autosomal recessive
LSM11	100 %	617910	?Aicardi-Goutieres syndrome 8, 619486 (3), Autosomal recessive
LSS	99.98 %	600909	Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive
LTA	100 %	153440	{Psoriatic arthritis, susceptibility to}, 607507 (3); {Myocardial infarction, susceptibility to}, 608446 (3); {Leprosy, susceptibility to, 4}, 610988 (3)
LTBP1	99.95 %	150390	Cutis laxa, autosomal recessive, type IIE, 619451 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LTBP2	99.97 %	602091	Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive
LTBP3	99.96 %	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant
LTBP4	99.99 %	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive
LTC4S	100 %	246530	Leukotriene C4 synthase deficiency, 614037 (1), Autosomal recessive
LTV1	99.92 %	620074	Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199 (3), Autosomal recessive
LYL1	100 %	151440	Leukemia, T-cell acute lymphoblastoid, 151440 (2)
LYN	99.95 %	165120	Autoinflammatory disease, systemic, with vasculitis, 620376 (3), Autosomal dominant
LYRM4	71.37 %	613311	?Combined oxidative phosphorylation deficiency 19, 615595 (3), Autosomal recessive
LYRM7	99.98 %	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LYZ	99.86 %	153450	Amyloidosis, hereditary systemic 5, 620658 (3)
LZTFL1	100 %	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
LZTR1	99.46 %	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
LZTS1	100 %	606551	Esophageal squamous cell carcinoma, somatic, 133239 (3)
M1AP	99.97 %	619098	Spermatogenic failure 48, 619108 (3), Autosomal recessive
MAB21L1	100 %	601280	Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive
MAB21L2	100 %	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal dominant, Autosomal recessive
MACF1	99.33 %	608271	Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant
MAD1L1	100 %	602686	Prostate cancer, somatic, 176807 (3); Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition, 620189 (3), Autosomal recessive; Lymphoma, B-cell, somatic (3)
MAD2L2	99.99 %	604094	?Fanconi anemia, complementation group V, 617243 (3), Autosomal recessive
MADD	99.95 %	603584	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive
MAF	99.73 %	177075	Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant
MAFA	99.98 %	610303	Insulinomatosis and diabetes mellitus, 147630 (3), Autosomal dominant
MAFB	100 %	608968	Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant
MAG	99.99 %	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
MAGED2	99.98 %	300470	Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive
MAGEL2	99.99 %	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
MAGI2	99.89 %	606382	Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive
MAGT1	99.54 %	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MAK	99.47 %	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
MALT1	99.71 %	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
MAML2	99.98 %	607537	Mucoepidermoid salivary gland carcinoma (3)
MAMLD1	99.84 %	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive
MAN1B1	99.94 %	604346	Rafiq syndrome, 614202 (3), Autosomal recessive
MAN2B1	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAN2C1	99.96 %	154580	Congenital disorder of deglycosylation 2, 619775 (3), Autosomal recessive
MANBA	99.81 %	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MANF	100 %	601916	Diabetes, deafness, developmental delay, and short stature syndrome, 620651 (3), Autosomal recessive
MAOA	99.83 %	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
MAP1B	100 %	157129	?Deafness, autosomal dominant 83, 619808 (3), Autosomal dominant; Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant
MAP2K1	99.98 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	99.99 %	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MAP3K1	99.93 %	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant
MAP3K14	99.98 %	604655	Immunodeficiency 112, 620449 (3), Autosomal recessive
MAP3K20	99.84 %	609479	Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive
MAP3K7	99.33 %	602614	Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant; Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant
MAP3K8	99.97 %	191195	Lung cancer, somatic, 211980 (3)
MAPK1	99.84 %	176948	Noonan syndrome 13, 619087 (3), Autosomal dominant
MAPK8IP1	100 %	604641	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
MAPK8IP3	100 %	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
MAPKAPK3	99.97 %	602130	?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant
MAPKAPK5	99.95 %	606723	Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive
MAPKBP1	99.98 %	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
MAPRE2	99.96 %	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
MAPT	99.8 %	157140	Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Frontotemporal dementia 1, with or without parkinsonism, 600274 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Pick disease, 172700 (3), Autosomal dominant
MARCHF6	99.97 %	613297	Epilepsy, familial adult myoclonic, 3, 613608 (3), Autosomal dominant
MARK3	99.88 %	602678	?Visual impairment and progressive phthisis bulbi, 618283 (3), Autosomal recessive
MARS1	99.97 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MARS2	100 %	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MARVELD2	99.72 %	610572	Deafness, autosomal recessive 49, 610153 (3), Autosomal recessive
MASP1	99.99 %	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MASP2	99.95 %	605102	MASP2 deficiency, 613791 (3), Autosomal recessive
MAST1	100 %	612256	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant
MAST3	97.04 %	612258	Developmental and epileptic encephalopathy 108, 620115 (3), Autosomal dominant
MAT1A	99.7 %	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal dominant, Autosomal recessive; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal dominant, Autosomal recessive
MATN3	99.97 %	602109	{Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant
MATR3	99.86 %	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
MAX	99.96 %	154950	Polydactyly-macrocephaly syndrome, 620712 (3), Autosomal dominant; {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
MB	99.99 %	160000	Myopathy, sarcoplasmic body, 620286 (3), Autosomal dominant
MBD4	99.98 %	603574	{Uveal melanoma, susceptibility to, 1}, 606660 (3), Autosomal dominant; Tumor predisposition syndrome 2, 619975 (3), Autosomal recessive
MBD5	99.79 %	611472	Intellectual developmental disorder, autosomal dominant 1, 156200 (3), Autosomal dominant
MBL2	99.93 %	154545	{Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant
MBOAT7	100 %	606048	Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive
MBTPS1	99.94 %	603355	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392 (3), Autosomal recessive
MBTPS2	99.81 %	300294	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MC1R	100 %	155555	[Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; {Melanoma, cutaneous malignant, 5}, 613099 (3); {Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive
MC2R	100 %	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
MC3R	100 %	155540	{Obesity, severe, susceptibility to, BMIQ9}, 602025 (3)
MC4R	100 %	155541	Obesity (BMIQ20), 618406 (3), Autosomal dominant, Autosomal recessive; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal dominant, Autosomal recessive
MCAT	99.99 %	614479	Optic atrophy 15, 620583 (3), Autosomal recessive
MCC	100 %	159350	Colorectal cancer, somatic, 114500 (3)
MCCC1	99.86 %	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
MCCC2	99.97 %	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
MCEE	99.9 %	608419	Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive
MCFD2	100 %	607788	Factor V and factor VIII, combined deficiency of, 613625 (3)
MCIDAS	100 %	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MCM10	99.99 %	609357	Immunodeficiency 80 with or without cardiomyopathy, 619313 (3), Autosomal recessive
MCM2	100 %	116945	?Deafness, autosomal dominant 70, 616968 (3), Autosomal dominant
MCM3AP	99.98 %	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
MCM4	99.96 %	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
MCM5	99.98 %	602696	?Meier-Gorlin syndrome 8, 617564 (3), Autosomal recessive
MCM6	99.79 %	601806	Lactase persistence/nonpersistence, 223100 (3), Autosomal dominant
MCM8	98.78 %	608187	?Premature ovarian failure 10, 612885 (3), Autosomal recessive
MCM9	99.76 %	610098	Ovarian dysgenesis 4, 616185 (3), Autosomal recessive
MCOLN1	100 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	100 %	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MCTS1	99.4 %	300587	Immunodeficiency 118, mycobacteriosis, 301115 (3), X-linked recessive
MDFIC	99.8 %	614511	Lymphatic malformation 12, 620014 (3), Autosomal recessive
MDH1	99.96 %	154200	?Developmental and epileptic encephalopathy 88, 618959 (3), Autosomal recessive
MDH2	99.54 %	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
MDM2	99.26 %	164785	{Accelerated tumor formation, susceptibility to}, 614401 (3), Autosomal dominant; ?Lessel-Kubisch syndrome, 618681 (3), Autosomal recessive
MDM4	99.43 %	602704	?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant
MECOM	99.97 %	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
MECP2	99.95 %	300005	Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MECR	99.63 %	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive; Optic atrophy 16, 620629 (3), Autosomal recessive
MED11	99.99 %	612383	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327 (3), Autosomal recessive
MED12	99.94 %	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MED12L	99.95 %	611318	Nizon-Isidor syndrome, 618872 (3), Autosomal dominant
MED13	99.55 %	603808	Intellectual developmental disorder, autosomal dominant 61, 618009 (3), Autosomal dominant
MED13L	99.99 %	608771	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant
MED17	99.82 %	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
MED23	99.82 %	605042	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 (3), Autosomal recessive
MED25	99.95 %	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive
MED27	99.99 %	605044	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MEF2A	99.98 %	600660	{Coronary artery disease, autosomal dominant, 1}, 608320 (3), Autosomal dominant
MEF2C	99.57 %	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 (3), Autosomal dominant
MEFV	100 %	608107	Neutrophilic dermatosis, acute febrile, 608068 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive; Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant
MEGF10	99.91 %	612453	Congenital myopathy 10A, severe variant, 614399 (3), Autosomal recessive; Congenital myopathy 10B, mild variant, 620249 (3), Autosomal recessive
MEGF8	99.9 %	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEI1	99.99 %	608797	Hydatidiform mole, recurrent, 3, 618431 (3), Autosomal recessive
MEIOB	99.98 %	617670	Premature ovarian failure 23, 620686 (3), Autosomal recessive; Spermatogenic failure 22, 617706 (3), Autosomal recessive
MEIS2	99.97 %	601740	Cleft palate, cardiac defects, and impaired intellectual development, 600987 (3), Autosomal dominant
MEN1	99.98 %	613733	Lipoma, somatic (3); Angiofibroma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Carcinoid tumor of lung (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3)
MEOX1	99.99 %	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
MERTK	98.9 %	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
MESD	99.98 %	607783	Osteogenesis imperfecta, type XX, 618644 (3), Autosomal recessive
MESP2	99.99 %	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive
MET	99.97 %	164860	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3); ?Arthrogryposis, distal, type 11, 620019 (3), Autosomal dominant; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive
METTL13	99.89 %	617987	{?Deafness, autosomal recessive 26, modifier of}, 605429 (3), Autosomal dominant
METTL23	100 %	615262	Intellectual developmental disorder, autosomal recessive 44, 615942 (3), Autosomal recessive
METTL5	99.75 %	618628	Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive
MFAP5	99.96 %	601103	Aortic aneurysm, familial thoracic 9, 616166 (3), Autosomal dominant
MFF	99.97 %	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
MFHAS1	100 %	605352	Malignant fibrous histiocytoma, 605352 (2)
MFN2	99.98 %	608507	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
MFRP	100 %	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
MFSD2A	99.78 %	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive
MFSD8	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MGAT2	100 %	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MGME1	100 %	615076	Mitochondrial DNA depletion syndrome 11, 615084 (3), Autosomal recessive
MGP	99.95 %	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MIA3	99.84 %	613455	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269 (3), Autosomal recessive
MIAT	81.85 %	611082	{Myocardial infarction, susceptibility to}, 608446 (3)
MIB1	99.91 %	608677	Left ventricular noncompaction 7, 615092 (3), Autosomal dominant
MICOS13	99.95 %	616658	Combined oxidative phosphorylation deficiency 37, 618329 (3), Autosomal recessive
MICU1	99.56 %	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MID1	99.9 %	300552	Opitz GBBB syndrome, 300000 (3), X-linked recessive
MID2	99.8 %	300204	?Intellectual developmental disorder, X-linked 101, 300928 (3), X-linked recessive
MIEF1	99.99 %	615497	Optic atrophy 14, 620550 (3), Autosomal dominant
MIEF2	100 %	615498	?Combined oxidative phosphorylation deficiency 49, 619024 (3), Autosomal recessive
MIF	100 %	153620	{Rheumatoid arthritis, systemic juvenile, susceptibility to}, 604302 (3)
MINAR2	99.99 %	620215	Deafness, autosomal recessive 120, 620238 (3), Autosomal recessive
MINPP1	99.73 %	605391	{Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive
MIP	99.98 %	154050	Cataract 15, multiple types, 615274 (3), Autosomal dominant
MIPEP	99.95 %	602241	Combined oxidative phosphorylation deficiency 31, 617228 (3), Autosomal recessive
MIR140	0 %	611894	Spondyloepiphyseal dysplasia, Nishimura type, 618618 (3), Autosomal dominant
MIR184	100 %	613146	EDICT syndrome, 614303 (3), Autosomal dominant
MIR204	100 %	610942	Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
MIR2861	100 %	613405	[Bone mineral density QTL 15], 613418 (3), Autosomal dominant, Autosomal recessive
MIR96	100 %	611606	Deafness, autosomal dominant 50, 613074 (3), Autosomal dominant
MITF	99.98 %	156845	Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; COMMAD syndrome, 617306 (3), Autosomal recessive
MKKS	100 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKRN3	100 %	603856	Precocious puberty, central, 2, 615346 (3), Autosomal dominant
MKS1	99.92 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MLC1	99.99 %	605908	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive
MLH1	99.64 %	120436	Lynch syndrome 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive
MLH3	99.98 %	604395	{Endometrial cancer, susceptibility to}, 608089 (3), Somatic mutation, Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Colorectal cancer, hereditary nonpolyposis, type 7, 614385 (3)
MLIP	99.71 %	614106	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138 (3), Autosomal recessive
MLLT10	96.7 %	602409	Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant
MLPH	100 %	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MLYCD	99.95 %	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MMAA	99.95 %	607481	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive
MMAB	99.99 %	607568	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive
MMACHC	99.98 %	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	99.76 %	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
MMD2	100 %	613318	Miyoshi muscular dystrophy 2, 613318 (2)
MME	97.17 %	120520	?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal dominant, Autosomal recessive
MMP13	99.96 %	600108	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant; Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive
MMP14	99.97 %	600754	Winchester syndrome, 277950 (3), Autosomal recessive
MMP19	99.98 %	601807	Cavitary optic disc anomalies, 611543 (3), Autosomal dominant
MMP2	99.96 %	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
MMP20	99.98 %	604629	Amelogenesis imperfecta, type IIA2, 612529 (3), Autosomal recessive
MMP21	99.99 %	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
MMP3	99.8 %	185250	{Coronary heart disease, susceptibility to, 6}, 614466 (3)
MMP9	100 %	120361	Metaphyseal anadysplasia 2, 613073 (3), Autosomal recessive
MMUT	99.68 %	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MN1	99.98 %	156100	CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant
MNS1	99.86 %	610766	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 (3), Autosomal recessive
MNX1	99.83 %	142994	Currarino syndrome, 176450 (3), Autosomal dominant
MOCOS	99.98 %	613274	Xanthinuria, type II, 603592 (3), Autosomal recessive
MOCS1	99.95 %	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
MOCS2	99.96 %	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
MOG	100 %	159465	?Narcolepsy 7, 614250 (3), Autosomal dominant
MOGS	100 %	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MORC2	100 %	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant; Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 (3), Autosomal dominant
MOS	100 %	190060	Oocyte/zygote/embryo maturation arrest 20, 620383 (3), Autosomal recessive
MOV10L1	99.96 %	605794	?Spermatogenic failure 73, 619878 (3), Autosomal recessive
MPC1	100 %	614738	Mitochondrial pyruvate carrier deficiency, 614741 (3), Autosomal recessive
MPDU1	99.97 %	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPDZ	99.86 %	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
MPEG1	100 %	610390	Immunodeficiency 77, 619223 (3), Autosomal dominant
MPI	99.95 %	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive
MPIG6B	100 %	606520	?Thrombocytopenia, anemia, and myelofibrosis, 617441 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MPL	99.96 %	159530	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Amegakaryocytic thrombocytopenia, congenital, 1, 604498 (3), Autosomal recessive; Thrombocythemia 2, 601977 (3), Somatic mutation, Autosomal dominant
MPLKIP	99.99 %	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MPO	99.97 %	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Myeloperoxidase deficiency, 254600 (3), Autosomal recessive; {Lung cancer, protection against, in smokers} (3)
MPV17	99.98 %	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
MPZ	99.99 %	159440	Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant
MPZL2	100 %	604873	Deafness, autosomal recessive 111, 618145 (3), Autosomal recessive
MRAP	100 %	609196	Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive
MRAP2	99.94 %	615410	{?Obesity, susceptibility to, BMIQ18}, 615457 (3), Autosomal dominant
MRAS	99.97 %	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
MRE11	99.93 %	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRM2	99.98 %	606906	Mitochondrial DNA depletion syndrome 17, 618567 (3), Autosomal recessive
MRPL12	100 %	602375	?Combined oxidative phosphorylation deficiency 45, 618951 (3), Autosomal recessive
MRPL3	99.94 %	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
MRPL39	99.83 %	611845	Combined oxidative phosphorylation deficiency 59, 620646 (3), Autosomal recessive
MRPL44	99.86 %	611849	Combined oxidative phosphorylation deficiency 16, 615395 (3), Autosomal recessive
MRPS14	99.99 %	611978	?Combined oxidative phosphorylation deficiency 38, 618378 (3), Autosomal recessive
MRPS16	99.99 %	609204	Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive
MRPS2	99.99 %	611971	Combined oxidative phosphorylation deficiency 36, 617950 (3), Autosomal recessive
MRPS22	99.87 %	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
MRPS23	100 %	611985	?Combined oxidative phosphorylation deficiency 46, 618952 (3), Autosomal recessive
MRPS25	99.97 %	611987	?Combined oxidative phosphorylation deficiency 50, 619025 (3), Autosomal recessive
MRPS28	99.99 %	611990	?Combined oxidative phosphorylation deficiency 47, 618958 (3), Autosomal recessive
MRPS34	100 %	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
MRPS7	100 %	611974	?Combined oxidative phosphorylation deficiency 34, 617872 (3), Autosomal recessive
MRTFA	92.99 %	606078	?Immunodeficiency 66, 618847 (3), Autosomal recessive
MS4A1	99.6 %	112210	?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MSH2	99.23 %	609309	Lynch syndrome 1, 120435 (3), Autosomal dominant; Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 2, 619096 (3), Autosomal recessive
MSH3	99.35 %	600887	Familial adenomatous polyposis 4, 617100 (3), Autosomal recessive; Endometrial carcinoma, somatic, 608089 (3)
MSH4	89.29 %	602105	Premature ovarian failure 20, 619938 (3), Autosomal recessive; Spermatogenic failure 2, 108420 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MSH5	100 %	603382	?Premature ovarian failure 13, 617442 (3), Autosomal recessive; Spermatogenic failure 74, 619937 (3), Autosomal recessive
MSH6	99.97 %	600678	Lynch syndrome 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Somatic mutation, Autosomal dominant
MSL3	99.83 %	300609	Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant
MSMB	99.86 %	157145	{Prostate cancer, hereditary, 13}, 611928 (3)
MSMO1	99.88 %	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
MSN	99.98 %	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
MSR1	99.99 %	153622	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
MSRB3	99.69 %	613719	Deafness, autosomal recessive 74, 613718 (3), Autosomal recessive
MST1R	99.99 %	600168	{Nasopharyngeal carcinoma, susceptibility to, 3}, 617075 (3), Autosomal dominant
MSTN	99.96 %	601788	?Muscle hypertrophy, 614160 (3), Autosomal recessive
MSTO1	76.34 %	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
MSX1	100 %	142983	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant; Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3), Autosomal dominant
MSX2	100 %	123101	Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant; Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina 1, 168500 (3), Autosomal dominant
MTAP	99.87 %	156540	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250 (3), Autosomal dominant
MTFMT	99.98 %	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
MTHFD1	100 %	172460	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive
MTHFR	99.97 %	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
MTHFS	100 %	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive
MTM1	99.76 %	300415	Myopathy, centronuclear, X-linked, 310400 (3), X-linked recessive
MTMR14	99.99 %	611089	{Centronuclear myopathy, autosomal, modifier of}, 160150 (3), Autosomal dominant
MTMR2	99.94 %	603557	Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive
MTNR1B	100 %	600804	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
MTO1	90.25 %	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
MTOR	99.98 %	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant
MTPAP	99.97 %	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
MTR	99.95 %	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
MTRFR	99.87 %	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MTRR	99.98 %	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTSS2	100 %	616951	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086 (3), Autosomal dominant
MTTP	99.92 %	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive
MTX2	99.2 %	608555	Mandibuloacral dysplasia progeroid syndrome, 619127 (3), Autosomal recessive
MUC1	99.98 %	158340	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000 (3), Autosomal dominant
MUC5B	99.94 %	600770	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500 (3), Autosomal dominant
MUC7	100 %	158375	{Asthma, protection against}, 600807 (3), Autosomal dominant
MUSK	99.93 %	601296	Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
MUTYH	99.94 %	604933	Adenomas, multiple colorectal, 608456 (3), Autosomal recessive; Gastric cancer, somatic, 613659 (3)
MVD	100 %	603236	Porokeratosis 7, multiple types, 614714 (3), Autosomal dominant
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MXI1	99.98 %	600020	Prostate cancer, somatic, 176807 (3); Neurofibrosarcoma, somatic (3)
MYB	99.89 %	189990	{T-cell acute lymphoblastic leukemia} (3)
MYBPC1	99.77 %	160794	Congenital myopathy 16, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive; Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant
MYBPC3	99.98 %	600958	Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
MYC	100 %	190080	Burkitt lymphoma, somatic, 113970 (3)
MYCN	100 %	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant; Megalencephaly-polydactyly syndrome, 620748 (3), Autosomal dominant
MYD88	99.99 %	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive
MYF5	99.99 %	159990	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155 (3), Autosomal recessive
MYH10	99.91 %	160776	*No OMIM phenotypes*
MYH11	99.16 %	160745	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant; Visceral myopathy 2, 619350 (3), Autosomal dominant
MYH14	99.98 %	608568	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant
MYH2	99.98 %	160740	Congenital myopathy 6 with ophthalmoplegia, 605637 (3), Autosomal dominant, Autosomal recessive
MYH3	99.99 %	160720	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant; Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 (3), Autosomal recessive; Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 (3), Autosomal dominant; Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MYH6	100 %	160710	{Sick sinus syndrome 3}, 614090 (3); Atrial septal defect 3, 614089 (3); Cardiomyopathy, dilated, 1EE, 613252 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant
MYH7	99.99 %	160760	Laing distal myopathy, 160500 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Congenital myopathy 7B, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Congenital myopathy 7A, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant
MYH8	100 %	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
MYH9	99.95 %	160775	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant; Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant
MYL1	99.75 %	160780	Congenital myopathy 14, 618414 (3), Autosomal recessive
MYL2	99.99 %	160781	Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant; Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424 (3), Autosomal recessive
MYL3	99.99 %	160790	Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal dominant, Autosomal recessive
MYL4	99.82 %	160770	?Atrial fibrillation, familial, 18, 617280 (3), Autosomal dominant
MYL9	100 %	609905	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365 (3), Autosomal recessive
MYLK	99.97 %	600922	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 7, 613780 (3), Autosomal dominant
MYLK2	100 %	606566	Cardiomyopathy, hypertrophic, 1, digenic, 192600 (3), Digenic dominant, Autosomal dominant
MYLPF	99.85 %	617378	Arthrogryposis, distal, type 1C, 619110 (3), Autosomal dominant, Autosomal recessive
MYMK	99.88 %	615345	Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive
MYMX	100 %	619912	?Carey-Fineman-Ziter syndrome 2, 619941 (3), Autosomal recessive
MYO15A	99.93 %	602666	Deafness, autosomal recessive 3, 600316 (3), Autosomal recessive
MYO18B	99.98 %	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive
MYO1E	99.94 %	601479	Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive
MYO1H	99.99 %	614636	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482 (3), Autosomal recessive
MYO3A	99.93 %	606808	Deafness, autosomal recessive 30, 607101 (3), Autosomal recessive; Deafness, autosomal dominant 90, 620722 (3), Autosomal dominant
MYO5A	99.94 %	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
MYO5B	100 %	606540	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 10, 619868 (3), Autosomal recessive
MYO6	99.69 %	600970	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive
MYO7A	99.96 %	276903	Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Usher syndrome, type 1B, 276900 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MYO9A	99.93 %	604875	Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive
MYO9B	99.97 %	602129	*No OMIM phenotypes*
MYOC	99.98 %	601652	Glaucoma 1A, primary open angle, 137750 (3), Autosomal dominant
MYOCD	99.99 %	606127	Megabladder, congenital, 618719 (3), Autosomal dominant
MYOD1	100 %	159970	Congenital myopathy 17, 618975 (3), Autosomal recessive
MYOF	99.91 %	604603	?Angioedema, hereditary, 7, 619366 (3), Autosomal dominant
MYORG	99.99 %	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
MYOT	99.93 %	604103	Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant
MYOZ2	99.99 %	605602	Cardiomyopathy, hypertrophic, 16, 613838 (3), Autosomal dominant
MYPN	99.88 %	608517	Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant; Congenital myopathy 24, 617336 (3), Autosomal recessive; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant
MYRF	99.98 %	608329	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant; Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant
MYSM1	94.16 %	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
MYT1	99.99 %	600379	*No OMIM phenotypes*
MYT1L	99.99 %	613084	Intellectual developmental disorder, autosomal dominant 39, 616521 (3), Autosomal dominant
MYZAP	100 %	614071	Cardiomyopathy, dilated, 2K, 620894 (3), Autosomal recessive
NAA10	99.99 %	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NAA15	99.71 %	608000	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant
NAA20	99.93 %	610833	Intellectual developmental disorder, autosomal recessive 73, 619717 (3), Autosomal recessive
NAA60	99.99 %	614246	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786 (3), Autosomal recessive
NAA80	100 %	607073	?Auroneurodental syndrome, 620830 (3), Autosomal recessive
NACC1	99.99 %	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
NADK2	99.91 %	615787	2,4-dienoyl-CoA reductase deficiency, 616034 (3), Autosomal recessive
NADSYN1	99.79 %	608285	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845 (3), Autosomal recessive
NAE1	99.81 %	603385	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210 (3), Autosomal recessive
NAF1	99.94 %	617868	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365 (3), Autosomal dominant
NAGA	100 %	104170	Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	100 %	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NAGS	99.99 %	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
NALCN	99.97 %	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NANOS1	99.99 %	608226	Spermatogenic failure 12, 615413 (3), Autosomal dominant
NANS	100 %	605202	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (3), Autosomal recessive
NAPB	100 %	611270	Developmental and epileptic encephalopathy 107, 620033 (3), Autosomal recessive
NARS1	99.94 %	108410	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 (3), Autosomal recessive
NARS2	99.59 %	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive
NAT2	100 %	612182	[Acetylation, slow], 243400 (3), Autosomal recessive
NAT8L	100 %	610647	?N-acetylaspartate deficiency, 614063 (3), Autosomal recessive
NAXD	99.99 %	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
NAXE	99.99 %	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
NBAS	99.86 %	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
NBEA	99.98 %	604889	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 (3), Autosomal dominant
NBEAL2	100 %	614169	Gray platelet syndrome, 139090 (3), Autosomal recessive
NBN	99.93 %	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCAPD2	100 %	615638	Microcephaly 21, primary, autosomal recessive, 617983 (3), Autosomal recessive
NCAPD3	99.98 %	609276	Microcephaly 22, primary, autosomal recessive, 617984 (3), Autosomal recessive
NCAPG2	99.97 %	608532	Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive
NCAPH	98.41 %	602332	?Microcephaly 23, primary, autosomal recessive, 617985 (3), Autosomal recessive
NCDN	99.99 %	608458	Neurodevelopmental disorder with infantile epileptic spasms, 619373 (3), Autosomal dominant
NCF1	57.22 %	608512	Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive
NCF2	99.85 %	608515	Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive
NCF4	100 %	601488	Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive
NCKAP1L	99.77 %	141180	Immunodeficiency 72 with autoinflammation, 618982 (3), Autosomal recessive
NCR3	100 %	611550	{Malaria, mild, susceptibility to}, 609148 (3)
NCSTN	99.82 %	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
NDE1	100 %	609449	Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive
NDNF	99.89 %	616506	Hypogonadotropic hypogonadism 25 with anosmia, 618841 (3), Autosomal dominant
NDP	99.98 %	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
NDRG1	99.99 %	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
NDST1	100 %	600853	Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive
NDUFA1	99.93 %	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
NDUFA10	99.98 %	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NDUFA11	98.22 %	612638	Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive
NDUFA12	99.21 %	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFA13	99.99 %	609435	{Thyroid carcinoma, Hurthle cell}, 607464 (3); Mitochondrial complex I deficiency, nuclear type 28, 618249 (3), Autosomal recessive
NDUFA2	99.95 %	602137	Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
NDUFA4	100 %	603833	?Mitochondrial complex IV deficiency, nuclear type 21, 619065 (3), Autosomal recessive
NDUFA6	99.96 %	602138	Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive
NDUFA8	100 %	603359	Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive
NDUFA9	100 %	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
NDUFAF1	100 %	606934	Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive
NDUFAF2	99.88 %	609653	Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive
NDUFAF3	100 %	612911	Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive
NDUFAF4	99.95 %	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
NDUFAF5	99.89 %	612360	Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive
NDUFAF6	99.86 %	612392	Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive
NDUFAF8	99.98 %	618461	Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive
NDUFB10	99.98 %	603843	?Mitochondrial complex I deficiency, nuclear type 35, 619003 (3), Autosomal recessive
NDUFB11	98.5 %	300403	Linear skin defects with multiple congenital anomalies 3, 300952 (3), X-linked dominant; ?Mitochondrial complex I deficiency, nuclear type 30, 301021 (3), X-linked
NDUFB3	99.6 %	603839	Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive
NDUFB7	99.89 %	603842	?Mitochondrial complex I deficiency, nuclear type 39, 620135 (3), Autosomal recessive
NDUFB8	99.99 %	602140	Mitochondrial complex I deficiency, nuclear type 32, 618252 (3), Autosomal recessive
NDUFB9	100 %	601445	?Mitochondrial complex I deficiency, nuclear type 24, 618245 (3), Autosomal recessive
NDUFC2	100 %	603845	Mitochondrial complex I deficiency, nuclear type 36, 619170 (3), Autosomal recessive
NDUFS1	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
NDUFS2	99.66 %	602985	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
NDUFS3	100 %	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
NDUFS4	99.99 %	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive
NDUFS6	100 %	603848	Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive
NDUFS7	99.99 %	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
NDUFS8	100 %	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
NDUFV1	99.99 %	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
NDUFV2	99.98 %	600532	Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive
NEB	87.33 %	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive; Arthrogryposis multiplex congenita 6, 619334 (3), Autosomal recessive
NECAP1	100 %	611623	Developmental and epileptic encephalopathy 21, 615833 (3), Autosomal recessive
NECTIN1	99.99 %	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NECTIN4	99.97 %	609607	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NEDD4L	99.97 %	606384	Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant
NEFH	100 %	162230	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant; {?Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal dominant, Autosomal recessive
NEFL	100 %	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
NEK1	99.83 %	604588	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Digenic recessive, Autosomal recessive; ?Orofaciodigital syndrome II, 252100 (3), Autosomal recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant
NEK10	96.92 %	618726	Ciliary dyskinesia, primary, 44, 618781 (3), Autosomal recessive
NEK2	95.09 %	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive
NEK8	99.99 %	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; Polycystic kidney disease 8, 620903 (3); ?Nephronophthisis 9, 613824 (3)
NEK9	99.96 %	609798	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3); Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive
NEMF	99.94 %	608378	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 (3), Autosomal recessive
NEPRO	99.9 %	617089	Anauxetic dysplasia 3, 618853 (3), Autosomal recessive
NEU1	99.98 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
NEUROD1	100 %	601724	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
NEUROD2	100 %	601725	Developmental and epileptic encephalopathy 72, 618374 (3), Autosomal dominant
NEUROG1	100 %	601726	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469 (3), Autosomal recessive
NEUROG3	100 %	604882	Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive
NEXMIF	99.99 %	300524	Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant
NEXN	97.73 %	613121	Cardiomyopathy, dilated, 1CC, 613122 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 20, 613876 (3), Autosomal dominant
NF1	99.88 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NF2	100 %	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, vestibular, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 101000 (3)
NFASC	99.94 %	609145	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive
NFATC2	100 %	600490	?Joint contracture, osteochondromas, and B-cell lymphoma, 620232 (3), Autosomal recessive
NFE2L2	99.97 %	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
NFIA	97.55 %	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NFIB	99.9 %	600728	Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant
NFIX	99.99 %	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant
NFKB1	99.8 %	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	99.98 %	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
NFKBIA	99.99 %	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant
NFKBIL1	100 %	601022	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
NFS1	100 %	603485	Combined oxidative phosphorylation deficiency 52, 619386 (3), Autosomal recessive
NFU1	99.48 %	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
NGF	100 %	162030	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive
NGLY1	99.93 %	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
NHEJ1	99.91 %	611290	Immunodeficiency 124, severe combined, 611291 (3), Autosomal recessive
NHLH2	100 %	162361	?Hypogonadotropic hypogonadism 27 without anosmia, 619755 (3), Autosomal recessive
NHLRC1	100 %	608072	Myoclonic epilepsy of Lafora 2, 620681 (3), Autosomal recessive
NHLRC2	99.83 %	618277	FINCA syndrome, 618278 (3), Autosomal recessive
NHP2	99.96 %	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NHS	99.96 %	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
NIN	99.82 %	608684	?Seckel syndrome 7, 614851 (3), Autosomal recessive
NIPA1	99.91 %	608145	Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant
NIPAL4	100 %	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
NIPBL	99.34 %	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKAP	99.1 %	300766	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive
NKX2-1	100 %	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NKX2-5	99.75 %	600584	Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant
NKX2-6	100 %	611770	Persistent truncus arteriosus, 217095 (3); Conotruncal heart malformations, 217095 (3)
NKX3-2	99.99 %	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive
NKX6-2	100 %	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
NLGN1	99.94 %	600568	{Autism, susceptibility to, 20}, 618830 (3), Autosomal dominant
NLGN3	99.98 %	300336	{Autism susceptibility, X-linked 1}, 300425 (3), X-linked
NLGN4X	99.98 %	300427	Intellectual developmental disorder, X-linked, 300495 (3), X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), X-linked
NLRC4	99.95 %	606831	?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant; Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NLRP1	95.26 %	606636	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal dominant, Autosomal recessive; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant
NLRP12	99.99 %	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
NLRP2	99.99 %	609364	Oocyte/zygote/embryo maturation arrest 18, 620332 (3), Autosomal recessive
NLRP3	100 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NLRP5	100 %	609658	Oocyte/zygote/embryo maturation arrest 19, 620333 (3), Autosomal recessive
NLRP7	99.99 %	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
NME5	99.81 %	603575	Ciliary dyskinesia, primary, 48, without situs inversus, 620032 (3), Autosomal recessive
NME8	99.83 %	607421	?Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NMNAT1	99.93 %	608700	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 (3), Autosomal recessive; Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
NMNAT2	99.79 %	608701	*No OMIM phenotypes*
NNMT	100 %	600008	Homocysteine plasma level, 600008 (2)
NNT	99.87 %	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
NOBOX	99.95 %	610934	Premature ovarian failure 5, 611548 (3), Autosomal dominant
NOD2	99.98 %	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
NODAL	99.98 %	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
NOG	100 %	602991	Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Brachydactyly, type B2, 611377 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant
NOL3	100 %	605235	?Myoclonus, familial, 1, 614937 (3), Autosomal dominant
NONO	99.94 %	300084	Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked
NOP10	99.99 %	606471	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400 (3), Autosomal dominant; ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425 (3), Autosomal recessive; ?Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOP56	99.99 %	614154	Spinocerebellar ataxia 36, 614153 (3), Autosomal dominant
NOS1AP	99.91 %	605551	Nephrotic syndrome, type 22, 619155 (3), Autosomal recessive
NOS2	96.11 %	163730	{Malaria, resistance to}, 611162 (3)
NOS3	93.73 %	163729	{Coronary artery spasm 1, susceptibility to} (3); {Hypertension, susceptibility to}, 145500 (3), Multifactorial; {Placental abruption} (3); {Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant; {Hypertension, pregnancy-induced}, 189800 (3), Autosomal dominant; {Ischemic stroke, susceptibility to}, 601367 (3), Multifactorial

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NOTCH1	99.98 %	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NOTCH2	99.03 %	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NOTCH2NLC	49.88 %	618025	Tremor, hereditary essential, 6, 618866 (3), Autosomal dominant; Oculopharyngodistal myopathy 3, 619473 (3), Autosomal dominant; Neuronal intranuclear inclusion disease, 603472 (3), Autosomal dominant
NOTCH3	99.99 %	600276	Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant
NOVA2	99.95 %	601991	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC1L1	97.47 %	608010	[Ezetimibe, nonresponse to], 617966 (3); [Low density lipoprotein cholesterol level QTL 7], 617966 (3)
NPC2	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	99.05 %	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	99.89 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NPHP4	99.98 %	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
NPHS1	99.97 %	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
NPHS2	99.87 %	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
NPM1	11.76 %	164040	Leukemia, acute myeloid, somatic, 601626 (3)
NPPA	100 %	108780	Atrial standstill 2, 615745 (3), Autosomal recessive; Atrial fibrillation, familial, 6, 612201 (3), Autosomal dominant
NPR2	99.99 %	108961	Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant; Acromesomelic dysplasia 1, Maroteaux type, 602875 (3), Autosomal recessive
NPR3	99.99 %	108962	Boudin-Mortier syndrome, 619543 (3), Autosomal recessive
NPRL2	100 %	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
NPRL3	99.99 %	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant
NPSR1	99.96 %	608595	{Asthma, susceptibility to, 2}, 608584 (3)
NPTX1	100 %	602367	Spinocerebellar ataxia 50, 620158 (3), Autosomal dominant
NQO1	100 %	125860	{Breast cancer, poor survival after chemotherapy for} (3); {Leukemia, post-chemotherapy, susceptibility to} (3); {Benzene toxicity, susceptibility to} (3)
NQO2	99.97 %	160998	{?Breast cancer susceptibility}, 114480 (1), Somatic mutation, Autosomal dominant
NR0B1	99.98 %	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
NR0B2	99.97 %	604630	Obesity, mild, early-onset, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive
NR1H4	99.49 %	603826	Cholestasis, progressive familial intrahepatic, 5, 617049 (3), Autosomal recessive
NR2E3	100 %	604485	Retinitis pigmentosa 37, 611131 (3), Autosomal dominant, Autosomal recessive; Enhanced S-cone syndrome, 268100 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NR2F1	99.99 %	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
NR2F2	100 %	107773	46XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NR3C1	99.93 %	138040	Glucocorticoid resistance, 615962 (3), Autosomal dominant
NR3C2	100 %	600983	Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)
NR4A2	99.97 %	601828	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 (3), Autosomal dominant
NR4A3	100 %	600542	Chondrosarcoma, extraskeletal myxoid, 612237 (3)
NR5A1	99.92 %	184757	46XX sex reversal 4, 617480 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant; Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant
NRAS	99.66 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NRCAM	99.82 %	601581	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive
NRG1	99.82 %	142445	{?Schizophrenia, susceptibility to}, 603013 (1)
NRIP1	100 %	602490	?Congenital anomalies of kidney and urinary tract 3, 618270 (3), Autosomal dominant
NRL	100 %	162080	Retinitis pigmentosa 27, 613750 (3), Autosomal dominant; Retinal degeneration, autosomal recessive, clumped pigment type (3)
NRROS	100 %	615322	Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive
NRXN1	99.98 %	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
NSD1	99.98 %	606681	Sotos syndrome, 117550 (3), Autosomal dominant
NSD2	99.87 %	602952	Rauch-Steindl syndrome, 619695 (3), Autosomal dominant
NSDHL	99.87 %	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
NSF	52.85 %	601633	Developmental and epileptic encephalopathy 96, 619340 (3), Autosomal dominant
NSMCE2	99.92 %	617246	Seckel syndrome 10, 617253 (3), Autosomal recessive
NSMCE3	100 %	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
NSMF	100 %	608137	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 (3), Autosomal dominant
NSRP1	99.96 %	616173	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive
NSUN2	99.96 %	610916	Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive
NSUN3	99.94 %	617491	Combined oxidative phosphorylation deficiency 48, 619012 (3), Autosomal recessive
NSUN6	99.95 %	617199	Intellectual developmental disorder, autosomal recessive 82, 620779 (3), Autosomal recessive
NT5C2	99.96 %	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NT5C3A	99.97 %	606224	Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3), Autosomal recessive
NT5E	99.92 %	129190	Calcification of joints and arteries, 211800 (3), Autosomal recessive
NTF4	100 %	162662	Glaucoma 1, open angle, 1O, 613100 (3)
NTHL1	99.99 %	602656	Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive
NTN1	99.99 %	601614	Mirror movements 4, 618264 (3), Autosomal dominant
NTNG2	99.98 %	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
NTRK1	99.86 %	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
NTRK2	99.9 %	600456	Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant
NUAK2	99.96 %	608131	?Anencephaly 2, 619452 (3), Autosomal recessive
NUBPL	99.62 %	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
NUDT15	99.98 %	615792	{Thiopurines, poor metabolism of, 2}, 616903 (3), Autosomal dominant
NUDT2	100 %	602852	Intellectual developmental disorder with or without peripheral neuropathy, 619844 (3), Autosomal recessive
NUP107	97.46 %	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
NUP133	99.45 %	607613	?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive; Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive
NUP155	99.52 %	606694	?Atrial fibrillation 15, 615770 (3), Autosomal recessive
NUP160	99.58 %	607614	?Nephrotic syndrome, type 19, 618178 (3), Autosomal recessive
NUP188	99.88 %	615587	Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive
NUP205	99.92 %	614352	?Nephrotic syndrome, type 13, 616893 (3), Autosomal recessive
NUP214	99.98 %	114350	Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive
NUP37	98.05 %	609264	?Microcephaly 24, primary, autosomal recessive, 618179 (3), Autosomal recessive
NUP54	99.95 %	607607	Dystonia 37, early-onset, with striatal lesions, 620427 (3), Autosomal recessive
NUP62	100 %	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
NUP85	99.98 %	170285	Nephrotic syndrome, type 17, 618176 (3), Autosomal recessive
NUP88	99.96 %	602552	Fetal akinesia deformation sequence 4, 618393 (3), Autosomal recessive
NUP93	99.87 %	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
NUS1	99.9 %	610463	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
NXN	99.92 %	612895	Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive
NYX	100 %	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
OAS1	99.96 %	164350	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042 (3), Autosomal dominant
OAT	90.17 %	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OBSCN	99.99 %	608616	{Rhabdomyolysis, susceptibility to, 1}, 620235 (3), Autosomal recessive
OBSL1	100 %	610991	3-M syndrome 2, 612921 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OCA2	99.6 %	611409	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive
OCLN	82.91 %	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OCRL	99.89 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
ODAD1	96.04 %	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
ODAD2	98.19 %	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
ODAD3	99.96 %	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
ODAD4	99.85 %	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
ODAPH	99.99 %	614829	Amelogenesis imperfecta, type IIA4, 614832 (3), Autosomal recessive
ODC1	99.99 %	165640	Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant
OFD1	99.68 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
OGDH	99.93 %	613022	Oxoglutarate dehydrogenase deficiency, 203740 (3), Autosomal recessive
OGDHL	99.95 %	617513	Yoon-Bellen neurodevelopmental syndrome, 619701 (3), Autosomal recessive
OGG1	100 %	601982	Renal cell carcinoma, clear cell, somatic, 144700 (3)
OGT	99.79 %	300255	Intellectual developmental disorder, X-linked 106, 300997 (3), X-linked recessive
OLR1	99.99 %	602601	{Myocardial infarction, susceptibility to}, 608446 (3)
OPA1	99.95 %	605290	Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive
OPA3	100 %	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
OPCML	99.99 %	600632	Ovarian cancer, somatic, 167000 (3)
OPHN1	99.92 %	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive
OPLAH	100 %	614243	5-oxoprolinase deficiency, 260005 (3), Autosomal dominant, Autosomal recessive
OPN1LW	71.75 %	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
OPN1MW	30.23 %	300821	Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive
OPN1SW	99.97 %	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant
OPTN	99.98 %	602432	Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435 (3), Autosomal dominant, Autosomal recessive; {Glaucoma, normal tension, susceptibility to}, 606657 (3)
OR2J3	100 %	615016	[C3HEX, ability to smell], 615082 (3), Autosomal dominant
ORAI1	99.63 %	610277	Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant
ORC1	99.7 %	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
ORC4	99.7 %	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
ORC6	99.82 %	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OSBPL2	100 %	606731	Deafness, autosomal dominant 67, 616340 (3), Autosomal dominant
OSGEP	100 %	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
OSMR	99.95 %	601743	Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant
OSTM1	99.56 %	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
OTC	99.42 %	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
OTOA	76.14 %	607038	Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive
OTOF	99.99 %	603681	Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive
OTOG	99.98 %	604487	Deafness, autosomal recessive 18B, 614945 (3), Autosomal recessive
OTOGL	99.07 %	614925	Deafness, autosomal recessive 84B, 614944 (3), Autosomal recessive
OTUD5	99.93 %	300713	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive
OTUD6B	99.92 %	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
OTUD7A	99.04 %	612024	Neurodevelopmental disorder with hypotonia and seizures, 620790 (3), Autosomal recessive
OTULIN	99.95 %	615712	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive; {Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986 (3), Autosomal dominant
OTX2	100 %	600037	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
OVOL2	100 %	616441	Corneal dystrophy, posterior polymorphous, 1, 122000 (3), Autosomal dominant
OXCT1	99.82 %	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive
OXGR1	100 %	606922	Nephrolithiasis, calcium oxalate, 2, with nephrocalcinosis, 620374 (3), Autosomal dominant
OXR1	99.96 %	605609	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive
P2RX2	100 %	600844	Deafness, autosomal dominant 41, 608224 (3), Autosomal dominant
P2RY12	99.99 %	600515	Bleeding disorder, platelet-type, 8, 609821 (3), Autosomal recessive
P3H1	99.89 %	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
P3H2	99.93 %	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
P4HA2	100 %	600608	Myopia 25, autosomal dominant, 617238 (3), Autosomal dominant
P4HB	99.99 %	176790	Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant
P4HTM	100 %	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive
PABPN1	99.99 %	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
PACS1	99.96 %	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PACS2	99.99 %	610423	Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant
PADI3	99.35 %	606755	Uncombable hair syndrome, 191480 (3), Autosomal recessive
PADI6	99.47 %	610363	Oocyte/zygote/embryo maturation arrest 16, 617234 (3), Autosomal recessive
PAFAH1B1	99.96 %	601545	Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant
PAH	99.96 %	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PAICS	99.99 %	172439	?Phosphoribosylaminoimidazole carboxylase deficiency, 619859 (3), Autosomal recessive
PAK1	99.99 %	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant
PAK2	99.83 %	605022	?Knobloch syndrome 2, 618458 (3), Autosomal dominant
PAK3	92.53 %	300142	Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive
PALB2	99.71 %	610355	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442 (3), Autosomal dominant; {Pancreatic cancer, susceptibility to, 3}, 613348 (3), Autosomal dominant; Fanconi anemia, complementation group N, 610832 (3)
PALLD	99.99 %	608092	{Pancreatic cancer, susceptibility to, 1}, 606856 (3), Autosomal dominant
PAM16	100 %	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive
PANK2	99.99 %	606157	Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PANK4	100 %	606162	?Cataract 49, 619593 (3), Autosomal dominant
PANX1	100 %	608420	Oocyte/zygote/embryo maturation arrest 7, 618550 (3), Autosomal dominant
PAPPA2	99.94 %	619485	Short stature, Dauber-Argente type, 619489 (3), Autosomal recessive
PAPSS2	99.91 %	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
PARK7	99.93 %	602533	Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive
PARN	99.75 %	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant
PARS2	99.99 %	612036	Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive
PATL2	99.99 %	614661	Oocyte/zygote/embryo maturation arrest 4, 617743 (3), Autosomal recessive
PAX1	100 %	167411	Otofaciocervical syndrome 2 with T-cell deficiency, 615560 (3), Autosomal recessive
PAX2	99.99 %	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PAX3	100 %	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal dominant, Autosomal recessive; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
PAX4	100 %	167413	{Diabetes mellitus, ketosis-prone, susceptibility to}, 612227 (3), Autosomal dominant, Autosomal recessive; Maturity-onset diabetes of the young, type IX, 612225 (3); Diabetes mellitus, type 2, 125853 (3), Autosomal dominant
PAX5	99.82 %	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PAX6	99.95 %	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; Microphthalmia/coloboma 12, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
PAX7	99.83 %	167410	Congenital myopathy 19, 618578 (3), Autosomal recessive; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
PAX8	99.99 %	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PAX9	99.99 %	167416	Tooth agenesis, selective, 3, 604625 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PBRM1	99.79 %	606083	?Renal cell carcinoma, clear cell, 144700 (3)
PBX1	99.88 %	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
PC	99.99 %	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCARE	100 %	613425	Retinitis pigmentosa 54, 613428 (3), Autosomal recessive
PCBD1	99.84 %	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
PCCA	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH12	100 %	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCDH15	99.71 %	605514	Usher syndrome, type 1D/F digenic, 601067 (3), Digenic recessive, Autosomal recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive
PCDH19	99.98 %	300460	Developmental and epileptic encephalopathy 9, 300088 (3), X-linked
PCDHGC4	100 %	606305	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 (3), Autosomal recessive
PCGF2	99.78 %	600346	Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant
PCK1	100 %	614168	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 (3), Autosomal recessive
PCK2	100 %	614095	PEPCK deficiency, mitochondrial, 261650 (1), Autosomal recessive
PCLO	99.41 %	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
PCNA	100 %	176740	?Ataxia-telangiectasia-like disorder 2, 615919 (3), Autosomal recessive
PCNT	99.97 %	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PCSK1	99.99 %	162150	{Obesity, susceptibility to, BMIQ12}, 612362 (3); Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955 (3), Autosomal recessive
PCSK9	99.99 %	607786	{Low density lipoprotein cholesterol level QTL 1}, 603776 (3), Autosomal dominant; Hypercholesterolemia, familial, 3, 603776 (3), Autosomal dominant
PCYT1A	100 %	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 5, 620680 (3), Autosomal recessive
PCYT2	100 %	602679	Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive
PDCD1	99.99 %	600244	{Multiple sclerosis, disease progression, modifier of}, 126200 (3), Multifactorial; {Systemic lupus erythematosus, susceptibility to, 2}, 605218 (3)
PDCD10	99.94 %	609118	Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant
PDCD6IP	99.9 %	608074	?Microcephaly 29, primary, autosomal recessive, 620047 (3), Autosomal recessive
PDE10A	87.37 %	610652	Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive
PDE11A	99.87 %	604961	Pigmented nodular adrenocortical disease, primary, 2, 610475 (3), Autosomal dominant
PDE1C	99.97 %	602987	?Deafness, autosomal dominant 74, 618140 (3), Autosomal dominant
PDE2A	99.95 %	602658	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive
PDE3A	99.97 %	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
PDE4D	99.89 %	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
PDE6A	99.98 %	180071	Retinitis pigmentosa 43, 613810 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PDE6B	100 %	180072	Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant
PDE6C	99.91 %	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
PDE6D	99.94 %	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive
PDE6G	100 %	180073	Retinitis pigmentosa 57, 613582 (3), Autosomal recessive
PDE6H	99.97 %	601190	Retinal cone dystrophy 3, 610024 (3), Autosomal dominant, Autosomal recessive; Achromatopsia 6, 610024 (3), Autosomal dominant, Autosomal recessive
PDE8B	99.98 %	603390	Pigmented nodular adrenocortical disease, primary, 3, 614190 (3); Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant
PDGFB	99.99 %	190040	Meningioma, SIS-related, 607174 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3)
PDGFRA	99.94 %	173490	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Isolated cases, Somatic mutation
PDGFRB	99.99 %	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant
PDGFRL	100 %	604584	Hepatocellular cancer, somatic, 114550 (3); Colorectal cancer, somatic, 114500 (3)
PDHA1	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHA2	100 %	179061	Spermatogenic failure 70, 619828 (3), Autosomal recessive
PDHB	99.94 %	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
PDHX	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PDK3	99.49 %	300906	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 (3), X-linked dominant
PDLIM4	100 %	603422	{Osteoporosis, susceptibility to}, 166710 (3), Autosomal dominant
PDP1	100 %	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
PDSS1	95.7 %	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	99.87 %	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
PDX1	100 %	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3)
PDXK	99.9 %	179020	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511 (3), Autosomal recessive
PDYN	100 %	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
PDZD7	98.09 %	612971	Deafness, autosomal recessive 57, 618003 (3), Autosomal recessive; {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive; Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive
PDZD8	99.99 %	614235	Intellectual developmental disorder with autism and dysmorphic facies, 620021 (3), Autosomal recessive
PEPD	99.98 %	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PER2	99.99 %	603426	?Advanced sleep phase syndrome, familial, 1, 604348 (3), Autosomal dominant
PER3	99.97 %	603427	?Advanced sleep phase syndrome, familial, 3, 616882 (3), Autosomal dominant
PERCC1	99.97 %	618656	Diarrhea 11, malabsorptive, congenital, 618662 (3), Autosomal recessive
PERP	100 %	609301	Erythrokeratodermia variabilis et progressiva 7, 619209 (3), Autosomal recessive; Olmsted syndrome 2, 619208 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PET100	99.98 %	614770	Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive
PET117	100 %	614771	?Mitochondrial complex IV deficiency, nuclear type 19, 619063 (3), Autosomal recessive
PEX1	98.8 %	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
PEX10	100 %	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	99.62 %	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
PEX12	100 %	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
PEX13	99.36 %	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	100 %	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	99.94 %	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
PEX19	99.25 %	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX2	100 %	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX26	100 %	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive
PEX3	99.85 %	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
PEX5	99.89 %	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX6	99.99 %	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
PEX7	99.72 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PFKL	99.99 %	171860	Hemolytic anemia due to phosphofructokinase deficiency (1)
PFKM	99.57 %	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
PFN1	74.59 %	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
PGAM2	100 %	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
PGAP1	99.56 %	611655	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive
PGAP2	99.99 %	615187	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207 (3), Autosomal recessive
PGAP3	99.97 %	611801	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716 (3), Autosomal recessive
PGK1	99.93 %	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM1	96.77 %	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
PGM2L1	99.82 %	611610	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191 (3), Autosomal recessive
PGM3	99.94 %	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PGR	99.97 %	607311	?Progesterone resistance, 264080 (2), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PHACTR1	100 %	608723	Developmental and epileptic encephalopathy 70, 618298 (3), Autosomal dominant
PHB	61.13 %	176705	{Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant
PHC1	94.38 %	602978	?Microcephaly 11, primary, autosomal recessive, 615414 (3), Autosomal recessive
PHEX	99.83 %	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
PHF21A	99.91 %	608325	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant
PHF6	99.16 %	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
PHF8	99.87 %	300560	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive
PHGDH	99.79 %	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHIP	99.63 %	612870	Chung-Jansen syndrome, 617991 (3), Autosomal dominant
PHKA1	99.84 %	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
PHKA2	99.92 %	300798	Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive
PHKB	99.69 %	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
PHKG2	99.86 %	172471	Glycogen storage disease IXc, 613027 (3), Autosomal recessive
PHLDB1	100 %	612834	Osteogenesis imperfecta, type XXIII, 620639 (3), Autosomal recessive
PHOX2A	100 %	602753	Fibrosis of extraocular muscles, congenital, 2, 602078 (3), Autosomal recessive
PHOX2B	99.98 %	603851	{Neuroblastoma, susceptibility to, 2}, 613013 (3); Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880 (3), Autosomal dominant
PHYH	100 %	602026	Refsum disease, 266500 (3), Autosomal recessive
PHYKPL	100 %	614683	[?Phosphohydroxylysinuria], 615011 (3), Autosomal recessive
PI4K2A	99.85 %	609763	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732 (3), Autosomal recessive
PI4KA	99.76 %	600286	Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PI4KB	99.9 %	602758	Deafness, autosomal dominant 87, 620281 (3), Autosomal dominant
PIBF1	99.9 %	607532	Joubert syndrome 33, 617767 (3), Autosomal recessive
PICALM	99.81 %	603025	Leukemia, acute myeloid, somatic, 601626 (3)
PIDD1	100 %	605247	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 (3), Autosomal recessive
PIEZO1	99.98 %	611184	[ER blood group system], 620207 (3), Autosomal recessive; Lymphatic malformation 6, 616843 (3), Autosomal recessive; Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant
PIEZO2	98.45 %	613629	Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PIGA	99.81 %	311770	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3), X-linked recessive
PIGB	99.92 %	604122	Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive
PIGC	100 %	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
PIGF	98.59 %	600153	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 (3), Autosomal recessive
PIGG	99.99 %	616918	[Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive
PIGH	100 %	600154	Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive
PIGK	92.56 %	605087	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive
PIGL	99.98 %	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGM	99.97 %	610273	Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive
PIGN	99.91 %	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
PIGO	99.99 %	614730	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 (3), Autosomal recessive
PIGP	99.87 %	605938	Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive
PIGQ	99.99 %	605754	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 (3), Autosomal recessive
PIGS	100 %	610271	Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive
PIGT	99.95 %	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
PIGU	99.98 %	608528	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive
PIGV	100 %	610274	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 (3), Autosomal recessive
PIGW	99.87 %	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
PIGY	99.99 %	610662	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809 (3), Autosomal recessive
PIK3C2A	99.92 %	603601	Oculoskeletodental syndrome, 618440 (3), Autosomal recessive
PIK3CA	99.74 %	171834	Hemifacial myohyperplasia, somatic, 606773 (3); CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3)
PIK3CD	99.99 %	602839	Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive
PIK3CG	99.72 %	601232	Immunodeficiency 97 with autoinflammation, 619802 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PIK3R1	99.86 %	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant
PIK3R2	99.95 %	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
PIK3R5	99.99 %	611317	Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive
PIKFYVE	99.81 %	609414	Corneal fleck dystrophy, 121850 (3), Autosomal dominant
PINK1	99.69 %	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
PIP5K1C	99.94 %	606102	Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive
PISD	100 %	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive
PITPNM3	99.98 %	608921	Cone-rod dystrophy 5, 600977 (3), Autosomal dominant
PITRM1	99.89 %	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
PITX1	100 %	602149	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3), Autosomal dominant
PITX2	99.98 %	601542	Ring dermoid of cornea, 180550 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant
PITX3	100 %	602669	Cataract 11, multiple types, 610623 (3), Autosomal dominant, Autosomal recessive; Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal dominant, Autosomal recessive
PJVK	99.6 %	610219	Deafness, autosomal recessive 59, 610220 (3), Autosomal recessive
PKD1	99.98 %	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PKD1L1	99.84 %	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
PKD2	99.91 %	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PKDCC	99.99 %	614150	Rhizomelic limb shortening with dysmorphic features, 618821 (3), Autosomal recessive
PKHD1	99.95 %	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
PKHD1L1	99.82 %	607843	Deafness, autosomal recessive 124, 620794 (3), Autosomal recessive
PKLR	99.84 %	609712	Adenosine triphosphate, elevated, of erythrocytes, 102900 (3), Autosomal dominant; Pyruvate kinase deficiency, 266200 (3), Autosomal recessive
PKP1	99.98 %	601975	Ectodermal dysplasia/skin fragility syndrome, 604536 (3), Autosomal recessive
PKP2	94.27 %	602861	Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant
PLA2G2A	99.57 %	172411	{?Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant
PLA2G4A	98.99 %	600522	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372 (3), Autosomal recessive
PLA2G5	99.91 %	601192	[Fleck retina, familial benign], 228980 (3), Autosomal recessive
PLA2G6	99.98 %	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
PLA2G7	99.78 %	601690	Platelet-activating factor acetylhydrolase deficiency, 614278 (3), Autosomal recessive
PLAA	99.79 %	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
PLAAT3	99.53 %	613867	Lipodystrophy, familial partial, type 9, 620683 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PLAG1	99.99 %	603026	Adenomas, salivary gland pleomorphic, somatic, 181030 (3); Silver-Russell syndrome 4, 618907 (3), Autosomal dominant
PLAU	100 %	191840	Quebec platelet disorder, 601709 (3), Autosomal dominant; {Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant
PLCB1	99.98 %	607120	Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive
PLCB2	100 %	604114	Platelet PLC beta-2 deficiency (1)
PLCB3	100 %	600230	Spondylometaphyseal dysplasia with corneal dystrophy, 618961 (3), Autosomal recessive
PLCB4	99.99 %	600810	Auriculocondylar syndrome 2B, 620458 (3), Autosomal recessive; Auriculocondylar syndrome 2A, 614669 (3), Autosomal dominant
PLCD1	99.98 %	602142	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal dominant, Autosomal recessive
PLCE1	99.98 %	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
PLCG1	100 %	172420	?Immune dysregulation, autoimmunity, and autoinflammation, 620514 (3), Autosomal dominant
PLCG2	99.99 %	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
PLCH1	99.99 %	612835	Holoprosencephaly 14, 619895 (3), Autosomal recessive
PLCZ1	99.83 %	608075	Spermatogenic failure 17, 617214 (3), Autosomal recessive
PLD1	99.82 %	602382	Cardiac valvular dysplasia 1, 212093 (3), Autosomal recessive
PLD3	99.99 %	615698	?Spinocerebellar ataxia 46, 617770 (3), Autosomal dominant
PLEC	100 %	601282	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 (3), Autosomal recessive; Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex 5A, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive
PLEKHG2	99.99 %	611893	Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive
PLEKHG5	99.99 %	611101	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive
PLEKHM1	99.77 %	611466	?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant
PLG	99.89 %	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PLIN1	99.99 %	170290	Lipodystrophy, familial partial, type 4, 613877 (3), Autosomal dominant
PLIN4	99.81 %	613247	Myopathy with rimmed ubiquitin-positive autophagic vacuolation, autosomal dominant, 601846 (3), Autosomal dominant
PLK4	99.89 %	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PLN	99.97 %	172405	Cardiomyopathy, dilated, 1P, 609909 (3); Cardiomyopathy, hypertrophic, 18, 613874 (3), Autosomal dominant
PLOD1	99.93 %	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
PLOD2	99.66 %	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
PLOD3	99.89 %	603066	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PLP1	99.98 %	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PLPBP	99.99 %	604436	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290 (3), Autosomal recessive
PLS1	99.83 %	602734	Deafness, autosomal dominant 76, 618787 (3), Autosomal dominant
PLS3	99.75 %	300131	Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant; Diaphragmatic hernia 5, X-linked, 306950 (3), X-linked
PLVAP	99.97 %	607647	Diarrhea 10, protein-losing enteropathy type, 618183 (3), Autosomal recessive
PLXNA1	100 %	601055	Dworschak-Punetha neurodevelopmental syndrome, 619955 (3), Autosomal recessive
PLXND1	99.98 %	604282	Congenital heart defects, multiple types, 9, 620294 (3), Autosomal recessive
PMFBP1	99.89 %	618085	Spermatogenic failure 31, 618112 (3), Autosomal recessive
PMM2	99.93 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMP2	99.93 %	170715	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279 (3), Autosomal dominant
PMP22	100 %	601097	Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive
PMPCA	99.99 %	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
PMPCB	99.94 %	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
PMS2	70.47 %	600259	Lynch syndrome 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
PMVK	99.6 %	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant
PNKD	100 %	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PNKP	100 %	605610	?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNLDC1	100 %	619529	Spermatogenic failure 57, 619528 (3), Autosomal recessive
PNLIP	99.81 %	246600	?Pancreatic lipase deficiency, 614338 (3), Autosomal recessive
PNP	100 %	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
PNPLA1	99.98 %	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive
PNPLA2	100 %	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
PNPLA6	99.99 %	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
PNPLA8	99.92 %	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
PNPO	99.9 %	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
PNPT1	99.56 %	610316	Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
POC1A	99.98 %	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POC1B	100 %	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
POF1B	99.34 %	300603	?Premature ovarian failure 2B, 300604 (3), X-linked recessive
POFUT1	100 %	607491	Dowling-Degos disease 2, 615327 (3), Autosomal dominant
POGLUT1	99.99 %	615618	Dowling-Degos disease 4, 615696 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive
POGZ	99.49 %	614787	White-Sutton syndrome, 616364 (3), Autosomal dominant
POLA1	99.57 %	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
POLD1	99.96 %	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; Immunodeficiency 120, 620836 (3), Autosomal recessive; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
POLD3	99.97 %	611415	Immunodeficiency 122, 620869 (3)
POLE	99.99 %	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
POLG	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLG2	99.51 %	604983	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive
POLH	99.85 %	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
POLR1A	99.93 %	616404	Leukodystrophy, hypomyelinating, 27, 620675 (3), Autosomal recessive; Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant
POLR1B	99.86 %	602000	Treacher-Collins syndrome 4, 618939 (3), Autosomal dominant
POLR1C	100 %	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR1D	100 %	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal dominant, Autosomal recessive
POLR2A	99.95 %	180660	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant
POLR3A	99.97 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	99.94 %	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
POLR3F	99.97 %	617455	?Immunodeficiency 101 (varicella zoster virus-specific), 619872 (3), Autosomal dominant
POLR3GL	83.89 %	617457	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234 (3), Autosomal recessive
POLR3K	100 %	606007	Leukodystrophy, hypomyelinating, 21, 619310 (3), Autosomal recessive
POLRMT	99.99 %	601778	Combined oxidative phosphorylation deficiency 55, 619743 (3), Autosomal dominant, Autosomal recessive
POMC	99.99 %	176830	{Obesity, early-onset, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
POMGNT1	99.69 %	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
POMGNT2	100 %	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
POMK	100 %	615247	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive
POMP	99.95 %	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
POMT1	99.96 %	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive
POMT2	99.98 %	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive
PON1	99.7 %	168820	{Coronary artery spasm 2, susceptibility to} (3); {Organophosphate poisoning, sensitivity to} (3); {Coronary artery disease, susceptibility to} (3); {Microvascular complications of diabetes 5}, 612633 (3)
PON2	99.05 %	602447	{Coronary artery disease, susceptibility to} (3)
POP1	99.94 %	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive
POPDC3	99.96 %	605824	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848 (3), Autosomal recessive
POR	99.98 %	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
PORCN	99.93 %	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
POT1	99.91 %	606478	Tumor predisposition syndrome 3, 615848 (3), Autosomal dominant; ?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368 (3), Autosomal recessive; ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367 (3), Autosomal dominant
POU1F1	99.98 %	173110	Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal dominant, Autosomal recessive
POU3F3	99.96 %	602480	Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant
POU3F4	100 %	300039	Deafness, X-linked 2, 304400 (3), X-linked recessive
POU4F1	99.71 %	601632	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352 (3), Autosomal dominant
POU4F3	99.99 %	602460	Deafness, autosomal dominant 15/52, 602459 (3), Autosomal dominant
POU6F2	94.47 %	609062	{Wilms tumor susceptibility-5}, 601583 (3), Somatic mutation, Autosomal dominant
PPA2	99.85 %	609988	?Sudden cardiac failure, alcohol-induced, 617223 (3), Autosomal recessive; Sudden cardiac failure, infantile, 617222 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PPARG	99.96 %	601487	{Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 604367 (3), Autosomal dominant; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant; [Obesity, resistance to] (3); Obesity, severe, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; Carotid intimal medial thickness 1, 609338 (3)
PPCS	99.52 %	609853	Cardiomyopathy, dilated, 2C, 618189 (3), Autosomal recessive
PPFIBP1	99.11 %	603141	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive
PPIB	100 %	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive
PPIL1	100 %	601301	Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive
PPIP5K2	99.69 %	611648	Deafness, autosomal recessive 100, 618422 (3), Autosomal recessive
PPM1D	99.81 %	605100	Breast cancer, somatic, 114480 (3); Jansen-de Vries syndrome, 617450 (3), Autosomal dominant
PPM1K	99.98 %	611065	Maple syrup urine disease, mild variant, 615135 (3), Autosomal recessive
PPOX	99.85 %	600923	Variegate porphyria, childhood-onset, 620483 (3), Autosomal recessive; Variegate porphyria, 176200 (3), Autosomal dominant
PPP1CB	99.89 %	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PPP1R12A	99.08 %	602021	Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant
PPP1R13L	99.99 %	607463	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519 (3), Autosomal recessive
PPP1R15B	99.92 %	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
PPP1R17	99.99 %	604088	{Hypercholesterolemia, susceptibility to}, 143890 (3), Autosomal dominant, Autosomal recessive
PPP1R21	99.92 %	618159	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive
PPP1R3A	99.97 %	600917	Insulin resistance, severe, digenic, 125853 (3), Autosomal dominant
PPP2CA	99.99 %	176915	Houge-Janssens syndrome 3, 618354 (3), Autosomal dominant
PPP2R1A	100 %	605983	Houge-Janssens syndrome 2, 616362 (3), Autosomal dominant
PPP2R1B	99.77 %	603113	Lung cancer, somatic, 211980 (3)
PPP2R2B	99.88 %	604325	Spinocerebellar ataxia 12, 604326 (3), Autosomal dominant
PPP2R3C	99.79 %	615902	Spermatogenic failure 36, 618420 (3), Autosomal dominant; Myoectodermal gonadal dysgenesis syndrome, 618419 (3), Autosomal recessive
PPP2R5D	99.99 %	601646	Houge-Janssens syndrome 1, 616355 (3), Autosomal dominant
PPP3CA	99.84 %	114105	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant
PPT1	97.48 %	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PQBP1	99.99 %	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRCC	99.99 %	179755	Renal cell carcinoma, papillary, 605074 (3)
PRCD	100 %	610598	Retinitis pigmentosa 36, 610599 (3)
PRDM10	100 %	618319	?Birt-Hogg-Dube syndrome 2, 620459 (3), Autosomal dominant
PRDM12	100 %	616458	Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PRDM13	99.99 %	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
PRDM16	99.99 %	605557	Left ventricular noncompaction 8, 615373 (3), Autosomal dominant; Cardiomyopathy, dilated, 1LL, 615373 (3), Autosomal dominant
PRDM5	99.76 %	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
PRDM6	99.99 %	616982	Patent ductus arteriosus 3, 617039 (3), Autosomal dominant
PRDM8	99.99 %	616639	?Epilepsy, progressive myoclonic, 10, 616640 (3), Autosomal recessive
PRDX1	98.81 %	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400 (3), Autosomal recessive
PRDX3	99.94 %	604769	Spinocerebellar ataxia, autosomal recessive 32, 619862 (3), Autosomal recessive; Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 (3), Autosomal dominant
PREPL	99.63 %	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
PRF1	100 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRG4	99.69 %	604283	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive
PRICKLE1	99.87 %	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRICKLE3	99.98 %	300111	{Leber hereditary optic neuropathy, modifier of}, 308905 (3), X-linked dominant
PRIM1	99.07 %	176635	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005 (3), Autosomal recessive
PRIMPOL	99.86 %	615421	Myopia 22, autosomal dominant, 615420 (3), Autosomal dominant
PRKACA	99.95 %	601639	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3); Cardioacrofacial dysplasia 1, 619142 (3), Autosomal dominant
PRKACB	87.33 %	176892	Cardioacrofacial dysplasia 2, 619143 (3), Somatic mosaicism, Autosomal dominant
PRKACG	100 %	176893	?Bleeding disorder, platelet-type, 19, 616176 (3), Autosomal recessive
PRKAG2	99.96 %	602743	Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), Autosomal dominant; Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant
PRKAG3	99.97 %	604976	[Skeletal muscle glycogen content and metabolism QTL], 619030 (3), Autosomal recessive
PRKAR1A	100 %	188830	Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant
PRKAR1B	100 %	176911	Marbach-Schaaf neurodevelopmental syndrome, 619680 (3), Autosomal dominant
PRKCA	99.98 %	176960	Pituitary tumor, invasive (3)
PRKCD	99.96 %	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
PRKCG	99.99 %	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
PRKCH	99.99 %	605437	{Cerebral infarction, susceptibility to}, 601367 (3), Multifactorial
PRKCSH	99.99 %	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
PRKD1	99.96 %	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PRKDC	99.93 %	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive
PRKG1	99.13 %	176894	Aortic aneurysm, familial thoracic 8, 615436 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PRKG2	99.75 %	601591	Spondylometaphyseal dysplasia, Pagnamenta type, 619638 (3), Autosomal recessive; Acromesomelic dysplasia 4, 619636 (3), Autosomal recessive
PRKN	99.99 %	602544	Adenocarcinoma of lung, somatic, 211980 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3)
PRKRA	99.94 %	603424	Dystonia 16, 612067 (3), Autosomal recessive
PRLR	100 %	176761	Multiple fibroadenomas of the breast, 615554 (3), Autosomal dominant; Hyperprolactinemia, 615555 (3), Autosomal dominant, Autosomal recessive
PRMT7	99.95 %	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PRNP	100 %	176640	Spongiform encephalopathy with neuropsychiatric features, 606688 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant; Huntington disease-like 1, 603218 (3), Autosomal dominant; Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant
PROC	99.98 %	612283	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive
PRODH	4.29 %	606810	{Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive
PROK2	98.65 %	607002	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant
PROKR2	100 %	607123	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant
PROM1	99.97 %	604365	Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod dystrophy 12, 612657 (3), Autosomal dominant, Autosomal recessive
PROP1	99.88 %	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive
PRORP	99.8 %	609947	Combined oxidative phosphorylation deficiency 54, 619737 (3), Autosomal recessive
PROS1	99.84 %	176880	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive; Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant
PROZ	100 %	176895	[Protein Z deficiency], 614024 (3)
PRPF3	99.73 %	607301	Retinitis pigmentosa 18, 601414 (3), Autosomal dominant
PRPF31	99.99 %	606419	Retinitis pigmentosa 11, 600138 (3), Autosomal dominant
PRPF4	99.96 %	607795	Retinitis pigmentosa 70, 615922 (3), Autosomal dominant
PRPF6	99.98 %	613979	Retinitis pigmentosa 60, 613983 (3), Autosomal dominant
PRPF8	99.98 %	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant
PRPH	99.98 %	170710	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal dominant, Autosomal recessive
PRPH2	99.98 %	179605	Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive; Leber congenital amaurosis 18, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PRPS1	99.95 %	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
PRR12	100 %	616633	Neuroocular syndrome, 619539 (3), Autosomal dominant
PRRT2	99.97 %	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant
PRRX1	99.56 %	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal dominant, Autosomal recessive
PRSS1	100 %	276000	Pancreatitis, hereditary, 167800 (3), Autosomal dominant
PRSS12	99.98 %	606709	Intellectual developmental disorder, autosomal recessive 1, 249500 (3), Autosomal recessive
PRSS2	99.99 %	601564	{Pancreatitis, chronic, protection against}, 167800 (3), Autosomal dominant
PRSS56	100 %	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
PRUNE1	99.85 %	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
PRX	99.99 %	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive
PSAP	99.94 %	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
PSAT1	99.98 %	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSEN1	100 %	104311	Pick disease, 172700 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant
PSEN2	99.97 %	600759	Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant
PSENEN	100 %	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMA6	99.99 %	602855	{Myocardial infarction, susceptibility to}, 608446 (3)
PSMB1	99.99 %	602017	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038 (3), Autosomal recessive
PSMB10	99.98 %	176847	Immunodeficiency 121 with autoinflammation, 620807 (3), Autosomal dominant; Proteasome-associated autoinflammatory syndrome 5, 619175 (3), Autosomal recessive
PSMB4	99.83 %	602177	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591 (3), Autosomal recessive
PSMB8	99.96 %	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
PSMB9	99.68 %	177045	Proteasome-associated autoinflammatory syndrome 6, 620796 (3), Autosomal dominant
PSMC1	69.18 %	602706	?Birk-Aharoni syndrome, 620071 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PSMC3	99.93 %	186852	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354 (3), Autosomal recessive
PSMC3IP	99.82 %	608665	Ovarian dysgenesis 3, 614324 (3), Autosomal recessive
PSMD12	99.95 %	604450	Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant
PSMG2	99.98 %	609702	?Proteasome-associated autoinflammatory syndrome 4, 619183 (3), Autosomal recessive
PSPH	99.09 %	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PSTPIP1	99.91 %	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
PTCD3	99.77 %	614918	Combined oxidative phosphorylation deficiency 51, 619057 (3), Autosomal recessive
PTCH1	99.99 %	601309	Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTCH2	99.92 %	603673	Medulloblastoma, somatic, 155255 (3); Basal cell carcinoma, somatic, 605462 (3)
PTCHD1	99.97 %	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
PTDSS1	99.95 %	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTEN	99.89 %	601728	{Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
PTF1A	100 %	607194	Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive
PTGDR	99.99 %	604687	{Asthma, susceptibility to, 1}, 607277 (3)
PTGER2	99.98 %	176804	{Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
PTGIS	100 %	601699	Hypertension, essential, 145500 (3), Multifactorial
PTH	99.98 %	168450	Hypoparathyroidism, familial isolated 1, 146200 (3), Autosomal dominant, Autosomal recessive
PTH1R	99.93 %	168468	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive
PTHLH	99.89 %	168470	Brachydactyly, type E2, 613382 (3), Autosomal dominant
PTPA	100 %	600756	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482 (3), Autosomal recessive
PTPN1	99.99 %	176885	{Insulin resistance, susceptibility to}, 125853 (3), Autosomal dominant
PTPN11	99.98 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTPN12	98.12 %	600079	Colon cancer, somatic, 114500 (3)
PTPN14	99.97 %	603155	Choanal atresia and lymphedema, 613611 (3), Autosomal recessive
PTPN22	95.23 %	600716	{Rheumatoid arthritis, susceptibility to}, 180300 (3); {Systemic lupus erythematosus susceptibility to}, 152700 (3), Autosomal dominant; {Diabetes, type 1, susceptibility to}, 222100 (3), Autosomal recessive
PTPN23	100 %	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
PTPRC	93.9 %	151460	Immunodeficiency 105, severe combined, 619924 (3), Autosomal recessive
PTPRF	99.92 %	179590	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PTPRJ	99.97 %	600925	Colon cancer, somatic, 114500 (3); Thrombocytopenia 10, 620484 (3), Autosomal recessive
PTPRO	99.91 %	600579	Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive
PTPRQ	99.06 %	603317	Deafness, autosomal dominant 73, 617663 (3), Autosomal dominant; Deafness, autosomal recessive 84A, 613391 (3), Autosomal recessive
PTRH2	99.99 %	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PTRHD1	100 %	617342	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747 (3), Autosomal recessive
PTS	99.93 %	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
PUF60	100 %	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PUM1	98.14 %	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant; Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719 (3), Autosomal dominant
PURA	100 %	600473	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 (3), Autosomal dominant
PUS1	100 %	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PUS3	100 %	616283	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive
PUS7	99.89 %	616261	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive
PXDN	100 %	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
PYCR1	99.99 %	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
PYCR2	99.95 %	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
PYGL	99.99 %	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
PYGM	99.96 %	608455	McArdle disease, 232600 (3), Autosomal recessive
PYROXD1	99.77 %	617220	Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive
QARS1	100 %	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
QDPR	99.92 %	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
QRICH1	100 %	617387	Ververi-Brady syndrome, 617982 (3), Autosomal dominant
QRICH2	100 %	618304	Spermatogenic failure 35, 618341 (3), Autosomal recessive
QRSL1	99.97 %	617209	Combined oxidative phosphorylation deficiency 40, 618835 (3), Autosomal recessive
RAB11B	100 %	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
RAB18	99.76 %	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB23	99.97 %	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAB27A	99.94 %	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAB28	99.97 %	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RAB32	99.99 %	612906	{Parkinson disease 26, autosomal dominant, susceptibility to}, 620923 (3), Autosomal dominant
RAB33B	100 %	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
RAB34	100 %	610917	Orofaciodigital syndrome XX, 620718 (3), Autosomal recessive
RAB39B	99.99 %	300774	Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RAB3GAP1	99.73 %	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	99.69 %	609275	Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RAB5IF	100 %	619960	?Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development syndrome 2, 616994 (3), Autosomal recessive
RAB7A	100 %	602298	Charcot-Marie-Tooth disease, type 2B, 600882 (3), Autosomal dominant
RABL3	99.95 %	618542	{?Pancreatic cancer, susceptibility to, 5}, 618680 (3), Autosomal dominant
RAC1	99.75 %	602048	Intellectual developmental disorder, autosomal dominant 48, 617751 (3), Autosomal dominant
RAC2	99.99 %	602049	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 (3), Autosomal dominant; ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3), Autosomal recessive; Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3), Autosomal dominant
RAC3	100 %	602050	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant
RACGAP1	99.31 %	604980	Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789 (3), Autosomal recessive
RAD21	99.91 %	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive
RAD50	99.75 %	604040	Nijmegen breakage syndrome-like disorder, 613078 (3), Autosomal recessive
RAD51	90.17 %	179617	Mirror movements 2, 614508 (3), Autosomal dominant; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant
RAD51C	99.02 %	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
RAD51D	100 %	602954	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291 (3)
RAD54B	99.75 %	604289	Colon cancer, somatic, 114500 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
RAD54L	99.3 %	603615	{Breast cancer, invasive ductal}, 114480 (3), Somatic mutation, Autosomal dominant; Adenocarcinoma, colonic, somatic (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
RAF1	99.97 %	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant
RAG1	100 %	179615	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3)
RAG2	100 %	179616	Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
RAI1	99.22 %	607642	Smith-Magenis syndrome, 182290 (3), Isolated cases, Autosomal dominant
RALA	99.69 %	179550	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant
RALGAPA1	99.73 %	608884	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive
RANBP2	99.37 %	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RAP1B	98.57 %	179530	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654 (3), Autosomal dominant
RAP1GDS1	99.8 %	179502	Alfadhel syndrome, 620655 (3), Autosomal recessive
RAPGEF2	99.84 %	609530	?Epilepsy, familial adult myoclonic, 7, 618075 (3), Autosomal dominant
RAPSN	99.97 %	601592	Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
RARA	99.98 %	180240	Leukemia, acute promyelocytic, 612376 (1)
RARB	99.99 %	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal dominant, Autosomal recessive
RARS1	99.76 %	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
RARS2	99.88 %	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RASA1	99.05 %	139150	Capillary malformation-arteriovenous malformation 1, 608354 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3)
RASGRP1	100 %	603962	Immunodeficiency 64, 618534 (3), Autosomal recessive
RASGRP2	100 %	605577	?Bleeding disorder, platelet-type, 18, 615888 (3), Autosomal recessive
RAX	100 %	601881	Microphthalmia, syndromic 16, 611038 (3), Autosomal recessive
RAX2	100 %	610362	Retinitis pigmentosa 95, 620102 (3), Autosomal recessive; Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3)
RB1	99.84 %	614041	Small cell cancer of the lung, somatic, 182280 (3); Bladder cancer, somatic, 109800 (3); Retinoblastoma, trilateral, 180200 (3), Somatic mutation, Autosomal dominant; Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Somatic mutation, Autosomal dominant
RB1CC1	99.9 %	606837	Breast cancer, somatic, 114480 (3)
RBBP8	99.91 %	604124	Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3)
RBCK1	100 %	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RBL2	99.44 %	180203	Brunet-Wagner neurodevelopmental syndrome, 619690 (3), Autosomal recessive
RBM10	99.98 %	300080	TARP syndrome, 311900 (3), X-linked recessive
RBM12	100 %	607179	{Schizophrenia 19, susceptibility to}, 617629 (3), Autosomal dominant
RBM20	99.99 %	613171	Cardiomyopathy, dilated, 1DD, 613172 (3), Autosomal dominant
RBM28	99.99 %	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBM8A	99.37 %	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
RBMX	99.64 %	300199	?Intellectual developmental disorder, X-linked syndromic, Gustavson type, 309555 (3), X-linked recessive; ?Intellectual developmental disorder, X-linked syndromic, Shashi type, 300238 (3), X-linked recessive
RBP3	99.99 %	180290	?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive
RBP4	99.99 %	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
RBPJ	99.96 %	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RC3H1	99.22 %	609424	?Immune dysregulation and systemic hyperinflammation syndrome, 618998 (3), Autosomal recessive
RCBTB1	99.99 %	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RD3	100 %	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RDH11	99.99 %	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive
RDH12	99.98 %	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal dominant, Autosomal recessive
RDH5	100 %	601617	Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
RDX	99.81 %	179410	Deafness, autosomal recessive 24, 611022 (3), Autosomal recessive
REC114	99.89 %	618421	Oocyte/zygote/embryo maturation arrest 10, 619176 (3), Autosomal recessive
RECQL	99.91 %	600537	RECON progeroid syndrome, 620370 (3), Autosomal recessive
RECQL4	100 %	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
REEP1	99.97 %	609139	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011 (3), Autosomal recessive; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751 (3), Autosomal dominant
REEP2	99.99 %	609347	Spastic paraplegia 72A, autosomal dominant, 615625 (3), Autosomal dominant; ?Spastic paraplegia 72B, autosomal recessive, 620606 (3), Autosomal recessive
REEP6	100 %	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
REL	96.99 %	164910	Immunodeficiency 92, 619652 (3), Autosomal recessive
RELA	99.99 %	164014	Autoinflammatory disease, familial, Behcet-like-3, 618287 (3), Autosomal dominant
RELB	99.97 %	604758	?Immunodeficiency 53, 617585 (3), Autosomal recessive
RELN	99.98 %	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
RELT	100 %	611211	Amelogenesis imperfecta, type IIIC, 618386 (3), Autosomal recessive
REN	99.85 %	179820	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; [Hyperproreninemia] (3); Tubulointerstitial kidney disease, autosomal dominant, 4, 613092 (3), Autosomal dominant
REPS1	99.93 %	614825	?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive
RERE	99.94 %	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
REST	99.99 %	600571	Deafness, autosomal dominant 27, 612431 (3), Autosomal dominant; {Wilms tumor 6, susceptibility to}, 616806 (3), Autosomal dominant; Fibromatosis, gingival, 5, 617626 (3), Autosomal dominant
RET	99.97 %	164761	{Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant
RETN	99.99 %	605565	{Hypertension, insulin resistance-related, susceptibility to}, 125853 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
RETREG1	99.99 %	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
RFC1	99.87 %	102579	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575 (3), Autosomal recessive
RFT1	99.79 %	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RFWD3	99.9 %	614151	?Fanconi anemia, complementation group W, 617784 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RFX5	99.88 %	601863	?MHC class II deficiency 5, 620818 (3), Autosomal recessive; MHC class II deficiency 3, 620816 (3), Autosomal recessive
RFX6	99.87 %	612659	Mitchell-Riley syndrome, 615710 (3), Autosomal recessive
RFX7	99.99 %	612660	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330 (3), Autosomal dominant
RFXANK	100 %	603200	MHC class II deficiency 2, 620815 (3), Autosomal recessive
RFXAP	99.98 %	601861	MHC class II deficiency 4, 620817 (3), Autosomal recessive
RGR	99.79 %	600342	Retinitis pigmentosa 44, 613769 (3)
RGS5	99.77 %	603276	[Blood pressure regulation QTL], 145500 (2), Multifactorial
RGS9	99.9 %	604067	Prolonged electroretinal response suppression 1, 608415 (3), Autosomal recessive
RGS9BP	100 %	607814	Prolonged electroretinal response suppression 2, 620344 (3), Autosomal recessive
RHAG	99.9 %	180297	Overhydrated hereditary stomatocytosis, 185000 (3), Autosomal dominant; Anemia, hemolytic, Rh-null, regulator type, 268150 (3), Autosomal recessive
RHBDF2	99.95 %	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant
RHCE	95.59 %	111700	Rh-null disease, amorph type, 617970 (3), Autosomal recessive
RHD	74.45 %	111680	{Hemolytic disease of fetus and newborn, RH-induced}, 619462 (3), Isolated cases; [Blood group, RH system], 111690 (3)
RHO	100 %	180380	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3), Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal dominant, Autosomal recessive; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive
RHOA	84.16 %	165390	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727 (3)
RHOBTB2	100 %	607352	Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant
RHOH	99.99 %	602037	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307 (3), Autosomal recessive
RIC1	99.91 %	610354	CATIFA syndrome, 618761 (3), Autosomal recessive
RILPL1	100 %	614092	Oculopharyngodistal myopathy 4, 619790 (3), Autosomal dominant
RIMS2	99.97 %	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
RIN2	99.99 %	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
RINT1	99.99 %	610089	Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive
RIPK1	99.93 %	603453	Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant
RIPK4	99.99 %	605706	CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive
RIPOR2	99.97 %	611410	Deafness, autosomal dominant 21, 607017 (3), Autosomal dominant; ?Deafness, autosomal recessive 104, 616515 (3), Autosomal recessive
RIPPLY2	99.97 %	609891	?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive
RIT1	99.78 %	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RLBP1	99.99 %	180090	Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive; Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
RLIM	99.93 %	300379	Tonne-Kalscheuer syndrome, 300978 (3), X-linked
RMND1	99.92 %	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RMRP	100 %	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
RNASEH1	99.99 %	604123	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479 (3), Autosomal recessive
RNASEH2A	99.95 %	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	99.94 %	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	99.99 %	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNASEL	99.62 %	180435	Prostate cancer 1, 601518 (3), Autosomal dominant
RNASET2	99.99 %	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
RNF113A	99.99 %	300951	Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked
RNF125	99.98 %	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
RNF13	99.81 %	609247	Developmental and epileptic encephalopathy 73, 618379 (3), Autosomal dominant
RNF139	100 %	603046	Renal cell carcinoma, 144700 (3)
RNF168	99.97 %	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
RNF170	99.9 %	614649	Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant; Spastic paraplegia 85, autosomal recessive, 619686 (3), Autosomal recessive
RNF2	97.8 %	608985	Luo-Schoch-Yamamoto syndrome, 619460 (3), Autosomal dominant
RNF212	100 %	612041	?Spermatogenic failure 62, 619673 (3), Autosomal recessive; Recombination rate QTL 1, 612042 (3)
RNF213	99.99 %	613768	{Moyamoya disease 2, susceptibility to}, 607151 (3), Autosomal dominant, Autosomal recessive
RNF216	99.99 %	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
RNF220	99.68 %	616136	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive
RNF31	100 %	612487	Immunodeficiency 115 with autoinflammation, 620632 (3), Autosomal recessive
RNF43	99.73 %	612482	Sessile serrated polyposis cancer syndrome, 617108 (3), Autosomal dominant
RNF6	100 %	604242	Esophageal carcinoma, somatic, 133239 (3)
RNH1	99.99 %	173320	{Encephalopathy, acute, infection-induced, susceptibility to, 12}, 620461 (3), Autosomal recessive
RNPC3	88.9 %	618016	Pituitary hormone deficiency, combined or isolated, 7, 618160 (3), Autosomal recessive
RNU4ATAC	99.95 %	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
RNU7-1	33.9 %	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
ROBO1	99.83 %	602430	Pituitary hormone deficiency, combined or isolated, 8, 620303 (3), Autosomal dominant; Neurooculorenal syndrome, 620305 (3), Autosomal recessive; ?Nystagmus 8, congenital, autosomal recessive, 257400 (3), Autosomal recessive
ROBO2	99.8 %	602431	Vesicoureteral reflux 2, 610878 (3), Autosomal dominant
ROBO3	99.99 %	608630	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313 (3), Autosomal recessive
ROBO4	100 %	607528	Aortic valve disease 3, 618496 (3), Autosomal dominant
ROGDI	99.98 %	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ROM1	100 %	180721	Retinitis pigmentosa 7, digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
ROR1	97.47 %	602336	?Deafness, autosomal recessive 108, 617654 (3), Autosomal recessive
ROR2	99.99 %	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RORA	99.97 %	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant
RORB	99.86 %	601972	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 (3), Autosomal dominant
RORC	99.42 %	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
RP1	99.98 %	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal dominant, Autosomal recessive
RP1L1	100 %	608581	Occult macular dystrophy, 613587 (3), Autosomal dominant; Retinitis pigmentosa 88, 618826 (3), Autosomal recessive
RP2	99.58 %	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
RP9	99.83 %	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant
RPA1	99.99 %	179835	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767 (3), Autosomal dominant
RPE65	98.99 %	180069	Retinitis pigmentosa 20, 613794 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Leber congenital amaurosis 2, 204100 (3), Autosomal recessive
RPGR	94.45 %	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3), X-linked; Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
RPGRIP1	99.95 %	605446	Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	96.35 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
RPIA	99.84 %	180430	Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive
RPL10	84.91 %	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive
RPL10L	100 %	619655	?Spermatogenic failure 63, 619689 (3), Autosomal recessive
RPL11	99.81 %	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant
RPL13	99.96 %	113703	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728 (3), Autosomal dominant
RPL15	31.77 %	604174	Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
RPL18	100 %	604179	?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant
RPL21	40.51 %	603636	Hypotrichosis 12, 615885 (3), Autosomal dominant
RPL26	30.55 %	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
RPL27	99.83 %	607526	?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant
RPL35	99.99 %	618315	?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant
RPL35A	97.55 %	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
RPL3L	99.99 %	617416	Cardiomyopathy, dilated, 2D, 619371 (3), Autosomal recessive
RPL5	28.81 %	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
RPS10	0 %	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
RPS14	100 %	130620	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 (3)
RPS15A	22.14 %	603674	?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RPS17	100 %	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
RPS19	100 %	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS23	100 %	603683	Brachycephaly, trichomegaly, and developmental delay, 617412 (3), Autosomal dominant
RPS24	91.48 %	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
RPS26	8.99 %	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
RPS27	27.45 %	603702	?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant
RPS28	100 %	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
RPS29	99.96 %	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
RPS6KA3	98.93 %	300075	Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant
RPS7	88.5 %	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
RPSA	0 %	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
RRAGC	99.16 %	608267	Long-Olsen-Distelmaier syndrome, 620609 (3), Autosomal dominant
RRAGD	99.93 %	608268	Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152 (3), Autosomal dominant
RRAS2	99.94 %	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
RRM1	99.91 %	180410	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647 (3), Autosomal dominant, Autosomal recessive
RRM2B	99.97 %	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
RRP7A	97.99 %	619449	?Microcephaly 28, primary, autosomal recessive, 619453 (3), Autosomal recessive
RS1	99.92 %	300839	Retinoschisis, 312700 (3), X-linked recessive
RSPH1	99.87 %	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
RSPH3	99.94 %	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
RSPH4A	99.95 %	612647	Ciliary dyskinesia, primary, 11, 612649 (3), Autosomal recessive
RSPH9	99.99 %	612648	Ciliary dyskinesia, primary, 12, 612650 (3), Autosomal recessive
RSPO1	99.99 %	609595	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive
RSPO2	99.99 %	610575	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 (3), Autosomal recessive; Tetraamelia syndrome 2, 618021 (3), Autosomal recessive
RSPO4	99.99 %	610573	Anonychia congenita, 206800 (3), Autosomal recessive
RSPRY1	99.73 %	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
RSRC1	99.95 %	613352	Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive
RTEL1	100 %	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RTN2	99.98 %	603183	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854 (3), Autosomal recessive; Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant
RTN4IP1	99.98 %	610502	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive
RTN4R	99.93 %	605566	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
RTTN	99.93 %	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
RUBCN	100 %	613516	Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
RUNX1	100 %	151385	Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant
RUNX2	100 %	600211	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, 119600 (3), Autosomal dominant
RUSC2	99.99 %	611053	Intellectual developmental disorder, autosomal recessive 61, 617773 (3), Autosomal recessive
RXYLT1	99.48 %	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
RYR1	99.97 %	180901	Congenital myopathy 1B, autosomal recessive, 255320 (3), Autosomal recessive; Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000 (3), Autosomal dominant; King-Denborough syndrome, 619542 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant
RYR2	99.94 %	180902	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant; Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 (3), Autosomal dominant
RYR3	99.98 %	180903	Congenital myopathy 20, 620310 (3), Autosomal recessive
S1PR2	100 %	605111	Deafness, autosomal recessive 68, 610419 (3), Autosomal recessive
SACS	99.97 %	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
SAG	99.98 %	181031	Retinitis pigmentosa 47, autosomal recessive, 613758 (3), Autosomal recessive; Retinitis pigmentosa 96, autosomal dominant, 620228 (3), Autosomal dominant; Oguchi disease-1, 258100 (3), Autosomal recessive
SALL1	100 %	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SALL2	100 %	602219	?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive
SALL4	100 %	607343	?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant
SAMD12	100 %	618073	Epilepsy, familial adult myoclonic, 1, 601068 (3), Autosomal dominant
SAMD7	99.99 %	620493	Macular dystrophy with or without cone dysfunction, 620762 (3), Autosomal recessive
SAMD9	99.93 %	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SAMD9L	99.95 %	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; ?Spinocerebellar ataxia 49, 619806 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SAMHD1	99.98 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SAR1B	99.59 %	607690	Chylomicron retention disease, 246700 (3), Autosomal recessive
SARDH	99.98 %	604455	[Sarcosinemia], 268900 (3), Autosomal recessive
SARS1	98.53 %	607529	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive
SARS2	99.99 %	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
SASH1	99.88 %	607955	Dyschromatosis universalis hereditaria 1, 127500 (3), Autosomal dominant; ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 (3), Autosomal recessive
SASH3	99.99 %	300441	Immunodeficiency 102, 301082 (3), X-linked recessive
SASS6	95.06 %	609321	Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive
SATB1	100 %	602075	den Hoed-de Boer-Voisin syndrome, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant
SATB2	99.96 %	608148	Glass syndrome, 612313 (3), Autosomal dominant
SBDS	99.93 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SBF1	99.99 %	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive
SBF2	99.77 %	607697	Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive
SC5D	99.97 %	602286	Lathosterolosis, 607330 (3), Autosomal recessive
SCAF4	99.91 %	616023	Fliedner-Zweier syndrome, 620511 (3), Autosomal dominant
SCAPER	99.75 %	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
SCARB1	99.99 %	601040	[High density lipoprotein cholesterol level QTL6], 610762 (3)
SCARB2	99.99 %	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCARF2	99.94 %	613619	Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive
SCD5	99.99 %	608370	?Deafness, autosomal dominant 79, 619086 (3), Autosomal dominant
SCGB3A2	100 %	606531	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant
SCLT1	95.17 %	611399	*No OMIM phenotypes*
SCN10A	99.99 %	604427	Episodic pain syndrome, familial, 2, 615551 (3), Autosomal dominant
SCN11A	99.94 %	604385	Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant
SCN1A	99.94 %	182389	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant
SCN1B	99.98 %	600235	Generalized epilepsy with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Developmental and epileptic encephalopathy 52, 617350 (3), Autosomal recessive; Cardiac conduction defect, nonspecific, 612838 (3); Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3)
SCN2A	99.86 %	182390	Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant
SCN2B	100 %	601327	Atrial fibrillation, familial, 14, 615378 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SCN3A	99.76 %	182391	Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant
SCN3B	100 %	608214	Atrial fibrillation, familial, 16, 613120 (3), Autosomal dominant; Brugada syndrome 7, 613120 (3), Autosomal dominant
SCN4A	99.98 %	603967	Paramyotonia congenita, 168300 (3), Autosomal dominant; Hyperkalemic periodic paralysis, 170500 (3), Autosomal dominant; Congenital myopathy 22B, severe fetal, 620369 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Congenital myopathy 22A, classic, 620351 (3), Autosomal recessive
SCN4B	100 %	608256	Atrial fibrillation, familial, 17, 611819 (3), Autosomal dominant; Long QT syndrome 10, 611819 (3), Autosomal dominant
SCN5A	100 %	600163	Ventricular fibrillation, familial, 1, 603829 (3); Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Long QT syndrome 3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; Brugada syndrome 1, 601144 (3), Autosomal dominant; Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive
SCN8A	99.77 %	600702	?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant
SCN9A	99.83 %	603415	Erythermalgia, primary, 133020 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive
SCNM1	99.47 %	608095	Orofaciodigital syndrome XIX, 620107 (3), Autosomal recessive
SCNN1A	100 %	600228	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant
SCNN1B	99.38 %	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant
SCNN1G	99.94 %	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant
SCO1	99.98 %	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
SCO2	100 %	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
SCP2	94.94 %	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive
SCUBE3	99.96 %	614708	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184 (3), Autosomal recessive
SCYL1	100 %	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SCYL2	99.21 %	616365	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SDC3	99.81 %	186357	{Obesity, association with}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive
SDCCAG8	100 %	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SDHA	99.98 %	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Pheochromocytoma/paraganglioma syndrome 5, 614165 (3), Autosomal dominant
SDHAF1	99.99 %	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
SDHAF2	99.96 %	613019	Pheochromocytoma/paraganglioma syndrome 2, 601650 (3), Autosomal dominant
SDHB	97.32 %	185470	Pheochromocytoma/paraganglioma syndrome 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)
SDHC	99.67 %	602413	Pheochromocytoma/paraganglioma syndrome 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant
SDHD	82.93 %	602690	Pheochromocytoma/paraganglioma syndrome 1, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive
SDR9C7	99.99 %	609769	Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive
SEC23A	99.94 %	610511	Craniolenticulosutural dysplasia, 607812 (3), Autosomal dominant, Autosomal recessive
SEC23B	99.93 %	610512	?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive
SEC24D	99.94 %	607186	Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive
SEC31A	99.86 %	610257	?Halperin-Birk syndrome, 618651 (3), Autosomal recessive
SEC61A1	99.99 %	609213	Immunodeficiency, common variable, 15, 620670 (3), Autosomal dominant; ?Neutropenia, severe congenital, 11, autosomal dominant, 620674 (3), Autosomal dominant; Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
SEC63	99.84 %	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
SECISBP2	99.99 %	607693	Thyroid hormone metabolism, abnormal, 1, 609698 (3), Autosomal recessive
SELENBP1	99.97 %	604188	Extraoral halitosis due to MTO deficiency, 618148 (3), Autosomal recessive
SELENOI	99.86 %	607915	Spastic paraplegia 81, autosomal recessive, 618768 (3), Autosomal recessive
SELENON	93.61 %	606210	Congenital myopathy 3 with rigid spine, 602771 (3), Autosomal recessive
SEMA3A	99.42 %	603961	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant
SEMA4A	99.81 %	607292	Retinitis pigmentosa 35, 610282 (3), Autosomal recessive; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive
SEMA6B	99.98 %	608873	Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant
SEMA7A	99.9 %	607961	?Cholestasis, progressive familial intrahepatic, 11, 619874 (3), Autosomal recessive; [Blood group, John-Milton-Hagen system], 614745 (3)
SEPSECS	99.78 %	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SEPTIN12	99.94 %	611562	Spermatogenic failure 10, 614822 (3), Autosomal dominant
SEPTIN9	99.99 %	604061	Amyotrophy, hereditary neuralgic, 162100 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SERAC1	99.9 %	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SERPINA1	100 %	107400	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive
SERPINA3	99.99 %	107280	Alpha-1-antichymotrypsin deficiency (3); Cerebrovascular disease, occlusive (3)
SERPINA6	100 %	122500	Corticosteroid-binding globulin deficiency, 611489 (3), Autosomal dominant, Autosomal recessive
SERPINA7	99.98 %	314200	[Thyroxine-binding globulin QTL], 300932 (3), X-linked
SERPINB6	100 %	173321	?Deafness, autosomal recessive 91, 613453 (3), Autosomal recessive
SERPINB7	99.93 %	603357	Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive
SERPINB8	99.98 %	601697	Peeling skin syndrome 5, 617115 (3), Autosomal recessive
SERPINC1	99.89 %	107300	Thrombophilia 7 due to antithrombin III deficiency, 613118 (3), Autosomal dominant, Autosomal recessive
SERPIND1	100 %	142360	Thrombophilia 10 due to heparin cofactor II deficiency, 612356 (3), Autosomal dominant
SERPINE1	99.9 %	173360	Plasminogen activator inhibitor-1 deficiency, 613329 (3), Autosomal dominant, Autosomal recessive; {Transcription of plasminogen activator inhibitor, modulator of} (3)
SERPINF1	100 %	172860	Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive
SERPINF2	99.99 %	613168	Alpha-2-plasmin inhibitor deficiency, 262850 (3), Autosomal recessive
SERPING1	100 %	606860	Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal dominant, Autosomal recessive; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
SERPINH1	100 %	600943	{Preterm premature rupture of the membranes, susceptibility to}, 610504 (3), Multifactorial; Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive
SERPINI1	99.98 %	602445	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant
SET	78.28 %	600960	Intellectual developmental disorder, autosomal dominant 58, 618106 (3), Autosomal dominant
SETBP1	100 %	611060	Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant
SETD1A	99.99 %	611052	Epilepsy, early-onset, 2, with or without developmental delay, 618832 (3), Autosomal dominant; Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 (3), Autosomal dominant
SETD1B	99.99 %	611055	Intellectual developmental disorder with seizures and language delay, 619000 (3), Autosomal dominant
SETD2	99.91 %	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 70, 620157 (3), Autosomal dominant; Rabin-Pappas syndrome, 620155 (3), Autosomal dominant
SETD5	99.99 %	615743	Intellectual developmental disorder, autosomal dominant 23, 615761 (3), Autosomal dominant
SETX	99.97 %	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SF3B1	99.4 %	605590	Myelodysplastic syndrome, somatic, 614286 (3)
SF3B2	99.98 %	605591	Craniofacial microsomia, 164210 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SF3B4	99.65 %	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
SFRP4	99.99 %	606570	Pyle disease, 265900 (3), Autosomal recessive
SFTPA1	99.99 %	178630	Interstitial lung disease 1, 619611 (3), Autosomal dominant, Autosomal recessive
SFTPA2	99.81 %	178642	Interstitial lung disease 2, 178500 (3), Autosomal dominant
SFTPB	99.99 %	178640	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive
SFTPC	99.99 %	178620	Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3), Autosomal dominant
SFXN4	99.97 %	615564	Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive
SGCA	100 %	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive
SGCB	99.95 %	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive
SGCD	100 %	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive
SGCE	93.12 %	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
SGCG	99.99 %	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive
SGMS2	99.97 %	611574	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550 (3), Autosomal dominant
SGO1	99.9 %	609168	Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive
SGPL1	99.95 %	603729	RENI syndrome, 617575 (3), Autosomal recessive
SGSH	100 %	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SH2B3	99.95 %	605093	Thrombocythemia, somatic, 187950 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3)
SH2D1A	98.98 %	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SH3BP2	100 %	602104	Cherubism, 118400 (3), Autosomal dominant
SH3GL1	100 %	601768	Leukemia, acute myeloid, 601626 (1), Somatic mutation, Autosomal dominant
SH3KBP1	99.95 %	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
SH3PXD2B	100 %	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
SH3TC2	100 %	608206	Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant
SHANK1	99.99 %	604999	*No OMIM phenotypes*
SHANK2	99.97 %	603290	{Autism susceptibility 17}, 613436 (3)
SHANK3	98.45 %	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant
SHARPIN	100 %	611885	Autoinflammation with episodic fever and immune dysregulation, 620795 (3), Autosomal recessive
SHH	100 %	600725	Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant
SHMT2	99.92 %	138450	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive
SHOC1	99.67 %	618038	Spermatogenic failure 75, 619949 (3), Autosomal recessive
SHOC2	99.96 %	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SHOX	92.7 %	400020	Short stature, idiopathic familial, 300582 (3); Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive; Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant
SHPK	100 %	605060	[Sedoheptulokinase deficiency], 617213 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SHQ1	99.81 %	613663	Neurodevelopmental disorder with dystonia and seizures, 619922 (3), Autosomal recessive; ?Dystonia 35, childhood-onset, 619921 (3), Autosomal recessive
SHROOM4	99.78 %	300579	*No OMIM phenotypes*
SI	99.73 %	609845	Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive
SIAE	99.94 %	610079	{Autoimmune disease, susceptibility to, 6}, 613551 (3)
SIAH1	100 %	602212	Buratti-Harel syndrome, 619314 (3), Autosomal dominant
SIGMAR1	99.99 %	601978	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726 (3), Autosomal recessive; ?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive
SIK1	3.83 %	605705	Developmental and epileptic encephalopathy 30, 616341 (3), Autosomal dominant
SIK3	99.98 %	614776	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162 (3), Autosomal recessive
SIL1	99.95 %	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIM1	99.98 %	603128	*No OMIM phenotypes*
SIN3A	99.97 %	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
SIPA1L3	99.97 %	616655	?Cataract 45, 616851 (3), Autosomal recessive
SIX1	100 %	601205	Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootic syndrome 3, 608389 (3), Autosomal dominant
SIX3	100 %	603714	Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant
SIX5	100 %	600963	Branchiootorenal syndrome 2, 610896 (3)
SIX6	100 %	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
SKI	99.98 %	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SKIV2L	99.98 %	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
SLC10A1	99.99 %	182396	Hypercholanemia, familial 2, 619256 (3), Autosomal recessive
SLC10A2	99.99 %	601295	?Bile acid malabsorption, primary, 1, 613291 (3), Autosomal recessive
SLC10A7	99.95 %	611459	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive
SLC11A1	99.99 %	600266	{Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (3); {Buruli ulcer, susceptibility to}, 610446 (3)
SLC11A2	99.68 %	600523	Anemia, hypochromic microcytic, with iron overload 1, 206100 (3), Autosomal recessive
SLC12A1	99.9 %	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
SLC12A2	99.55 %	600840	Kilquist syndrome, 619080 (3), Autosomal recessive; Delpire-McNeill syndrome, 619083 (3), Autosomal dominant; Deafness, autosomal dominant 78, 619081 (3), Autosomal dominant
SLC12A3	99.84 %	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
SLC12A5	99.99 %	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive
SLC12A6	99.98 %	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2II, 620068 (3), Autosomal dominant
SLC13A3	99.98 %	606411	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive
SLC13A5	99.99 %	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive
SLC14A1	99.98 %	613868	[Blood group, Kidd], 111000 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC16A1	99.26 %	600682	Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal dominant, Autosomal recessive
SLC16A12	100 %	611910	Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant
SLC16A2	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC17A3	99.99 %	611034	[Uric acid concentration, serum, QTL4], 612671 (3), Autosomal dominant; {Gout susceptibility 4}, 612671 (3), Autosomal dominant
SLC17A5	99.71 %	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC17A8	99.74 %	607557	Deafness, autosomal dominant 25, 605583 (3), Autosomal dominant
SLC17A9	99.96 %	612107	Porokeratosis 8, disseminated superficial actinic type, 616063 (3), Autosomal dominant
SLC18A2	100 %	193001	Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive
SLC18A3	99.99 %	600336	Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive
SLC19A1	99.99 %	600424	Immunodeficiency 114, folate-responsive, 620603 (3), Autosomal recessive; ?Megaloblastic anemia, folate-responsive, 601775 (3), Autosomal recessive
SLC19A2	98.86 %	603941	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive
SLC19A3	99.95 %	606152	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 (3), Autosomal recessive
SLC1A1	99.98 %	133550	Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3)
SLC1A2	99.95 %	600300	Developmental and epileptic encephalopathy 41, 617105 (3), Autosomal dominant
SLC1A3	99.98 %	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
SLC1A4	99.97 %	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
SLC20A1	99.97 %	137570	*No OMIM phenotypes*
SLC20A2	99.95 %	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
SLC22A12	99.99 %	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
SLC22A18	99.95 %	602631	Breast cancer, somatic, 114480 (3); Lung cancer, somatic, 211980 (3); Rhabdomyosarcoma, somatic, 268210 (3)
SLC22A4	99.3 %	604190	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
SLC22A5	99.99 %	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive
SLC24A1	99.92 %	603617	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3), Autosomal recessive
SLC24A4	99.93 %	609840	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3), Autosomal recessive; Amelogenesis imperfecta, type IIA5, 615887 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blue/green eyes], 210750 (3), Autosomal recessive
SLC24A5	99.99 %	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive
SLC25A1	99.93 %	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive
SLC25A10	99.97 %	606794	?Mitochondrial DNA depletion syndrome 19, 618972 (3), Autosomal recessive
SLC25A11	100 %	604165	Pheochromocytoma/paraganglioma syndrome 6, 618464 (3), Autosomal dominant
SLC25A12	99.72 %	603667	Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC25A13	99.67 %	603859	Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive
SLC25A15	100 %	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A19	99.99 %	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
SLC25A20	100 %	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC25A21	99.98 %	607571	?Mitochondrial DNA depletion syndrome 18, 618811 (3), Autosomal recessive
SLC25A22	100 %	609302	Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive
SLC25A24	93 %	608744	Fontaine progeroid syndrome, 612289 (3), Autosomal dominant
SLC25A26	99.76 %	611037	Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive
SLC25A3	99.79 %	600370	Mitochondrial phosphate carrier deficiency, 610773 (3), Autosomal recessive
SLC25A32	99.98 %	138480	?Exercise intolerance, riboflavin-responsive, 616839 (3), Autosomal recessive
SLC25A36	99.81 %	616149	Hyperinsulinemic hypoglycemia, familial, 8, 620211 (3), Autosomal recessive
SLC25A38	99.98 %	610819	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive
SLC25A4	100 %	103220	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant
SLC25A42	99.99 %	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
SLC25A46	99.88 %	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
SLC26A1	100 %	610130	?Hypersulfaturia, 620372 (3), Autosomal recessive; ?Nephrolithiasis, calcium oxalate, 1, 167030 (3), Autosomal recessive
SLC26A2	100 %	606718	Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive
SLC26A3	99.97 %	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
SLC26A4	99.98 %	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive
SLC26A5	99.96 %	604943	?Deafness, autosomal recessive 61, 613865 (3), Autosomal recessive
SLC26A8	99.98 %	608480	Spermatogenic failure 3, 606766 (3), Autosomal dominant, Autosomal recessive
SLC27A4	100 %	604194	Ichthyosis prematurity syndrome, 608649 (3), Autosomal recessive
SLC28A1	99.98 %	606207	[Uridine-cytidineuria], 618477 (3), Autosomal recessive
SLC29A3	99.98 %	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC2A1	99.93 %	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant
SLC2A10	100 %	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC2A2	99.96 %	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
SLC2A9	99.98 %	606142	{Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal dominant, Autosomal recessive; Hypouricemia, renal, 2, 612076 (3), Autosomal dominant, Autosomal recessive
SLC30A10	99.99 %	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
SLC30A2	99.94 %	609617	Zinc deficiency, transient neonatal, 608118 (3), Autosomal dominant
SLC30A7	94.71 %	611149	Ziegler-Huang syndrome, 620501 (3), Autosomal recessive
SLC30A8	99.99 %	611145	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
SLC30A9	99.75 %	604604	Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive
SLC31A1	99.95 %	603085	Neurodegeneration and seizures due to copper transport defect, 620306 (3), Autosomal recessive
SLC32A1	100 %	616440	Generalized epilepsy with febrile seizures plus, type 12, 620755 (3), Autosomal dominant; Developmental and epileptic encephalopathy 114, 620774 (3), Autosomal dominant
SLC33A1	99.67 %	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Huppke-Brendel syndrome, 614482 (3), Autosomal recessive
SLC34A1	99.99 %	182309	?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant
SLC34A2	99.99 %	604217	Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive
SLC34A3	100 %	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
SLC35A1	99.81 %	605634	Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive
SLC35A2	99.97 %	314375	Congenital disorder of glycosylation, type IIm, 300896 (3), Somatic mosaicism, X-linked dominant
SLC35A3	94.67 %	605632	Arthrogryposis, impaired intellectual development, and seizures, 615553 (3), Autosomal recessive
SLC35B2	100 %	610788	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 (3), Autosomal recessive
SLC35C1	100 %	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC35D1	87.5 %	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive
SLC36A2	99.99 %	608331	[Iminoglycinuria], 242600 (3), Digenic recessive, Autosomal recessive; [Hyperglycinuria], 138500 (3), Autosomal dominant
SLC37A4	99.9 %	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIw, 619525 (3), Autosomal dominant; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
SLC38A3	100 %	604437	Developmental and epileptic encephalopathy 102, 619881 (3), Autosomal recessive
SLC38A8	99.99 %	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
SLC39A13	99.98 %	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive
SLC39A14	92.87 %	608736	?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive
SLC39A4	100 %	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
SLC39A5	99.99 %	608730	Myopia 24, autosomal dominant, 615946 (3), Autosomal dominant
SLC39A7	100 %	601416	Agammaglobulinemia 9, autosomal recessive, 619693 (3), Autosomal recessive
SLC39A8	99.95 %	608732	Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC3A1	99.98 %	104614	Cystinuria, 220100 (3), Autosomal dominant, Autosomal recessive
SLC40A1	99.7 %	604653	Hemochromatosis, type 4, 606069 (3), Autosomal dominant
SLC41A1	99.98 %	610801	?Nephronophthisis-like nephropathy 2, 619468 (3), Autosomal recessive
SLC44A1	99.9 %	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
SLC44A4	100 %	606107	?Deafness, autosomal dominant 72, 617606 (3), Autosomal dominant
SLC45A1	99.99 %	605763	Intellectual developmental disorder with neuropsychiatric features, 617532 (3), Autosomal recessive
SLC45A2	100 %	606202	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; Albinism, oculocutaneous, type IV, 606574 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive
SLC46A1	100 %	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC4A1	99.94 %	109270	[Blood group, Swann], 601550 (3); [Blood group, Wright], 112050 (3); Distal renal tubular acidosis 1, 179800 (3), Autosomal dominant; [Blood group, Waldner], 112010 (3); Spherocytosis, type 4, 612653 (3), Autosomal dominant; [Blood group, Froese], 601551 (3); Distal renal tubular acidosis 4 with hemolytic anemia, 611590 (3), Autosomal recessive; {Malaria, resistance to}, 611162 (3); Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Blood group, Diego], 110500 (3)
SLC4A10	99.69 %	605556	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746 (3), Autosomal recessive
SLC4A11	100 %	610206	Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive; Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive
SLC4A2	100 %	109280	?Osteopetrosis, autosomal recessive 9, 620366 (3), Autosomal recessive
SLC4A3	99.99 %	106195	Short QT syndrome 7, 620231 (3), Autosomal dominant
SLC4A4	99.97 %	603345	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 (3), Autosomal recessive
SLC51A	100 %	612084	?Cholestasis, progressive familial intrahepatic, 6, 619484 (3), Autosomal recessive
SLC51B	100 %	612085	?Bile acid malabsorption, primary, 2, 619481 (3), Autosomal recessive
SLC52A1	100 %	607883	Riboflavin deficiency, 615026 (3), Autosomal dominant
SLC52A2	100 %	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SLC52A3	99.94 %	613350	?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive
SLC5A1	99.98 %	182380	Glucose/galactose malabsorption, 606824 (3), Autosomal recessive
SLC5A2	99.99 %	182381	Renal glucosuria, 233100 (3), Autosomal dominant, Autosomal recessive
SLC5A5	99.75 %	601843	Thyroid dyshormonogenesis 1, 274400 (3), Autosomal recessive
SLC5A6	100 %	604024	Sodium-dependent multivitamin transporter deficiency, 618973 (3), Autosomal recessive; Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 (3), Autosomal recessive
SLC5A7	99.57 %	608761	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
SLC6A1	99.96 %	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
SLC6A17	99.79 %	610299	Intellectual developmental disorder, autosomal recessive 48, 616269 (3), Autosomal recessive
SLC6A19	99.99 %	608893	Hartnup disorder, 234500 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC6A2	99.97 %	163970	?Orthostatic intolerance, 604715 (3), Autosomal dominant
SLC6A3	99.96 %	126455	Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3)
SLC6A4	100 %	182138	{Obsessive-compulsive disorder}, 164230 (3), Autosomal dominant; {Anxiety-related personality traits}, 607834 (3)
SLC6A5	99.97 %	604159	Hyperekplexia 3, 614618 (3), Autosomal dominant, Autosomal recessive
SLC6A6	99.98 %	186854	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 (3), Autosomal recessive
SLC6A8	99.99 %	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SLC6A9	99.93 %	601019	Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive
SLC7A14	99.99 %	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
SLC7A6OS	99.96 %	619192	Epilepsy, progressive myoclonic, 12, 619191 (3), Autosomal recessive
SLC7A7	99.99 %	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC7A9	99.97 %	604144	Cystinuria, 220100 (3), Autosomal dominant, Autosomal recessive
SLC9A1	99.96 %	107310	Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive
SLC9A3	100 %	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
SLC9A3R1	100 %	604990	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant
SLC9A6	99.42 %	300231	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked
SLC9A7	99.91 %	300368	Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive
SLC9A9	99.85 %	608396	{?Autism susceptibility 16}, 613410 (3)
SLCO1B1	98.69 %	604843	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
SLCO1B3	99.85 %	605495	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
SLCO2A1	99.99 %	601460	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 (3), Autosomal dominant; PHOAR2-enteropathy syndrome, 614441 (3), Autosomal recessive
SLF2	99.89 %	610348	Atelis syndrome 1, 620184 (3), Autosomal recessive
SLFN14	99.99 %	614958	Bleeding disorder, platelet-type, 20, 616913 (3), Autosomal dominant
SLITRK1	100 %	609678	Tourette syndrome, 137580 (3), Autosomal dominant; ?Trichotillomania, 613229 (3), Autosomal dominant, Multifactorial
SLITRK2	100 %	300561	Intellectual developmental disorder, X-linked 111, 301107 (3), X-linked
SLITRK6	100 %	609681	Deafness and myopia, 221200 (3), Autosomal recessive
SLURP1	100 %	606119	Meleda disease, 248300 (3), Autosomal recessive
SLX4	100 %	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMAD2	99.92 %	601366	Loeys-Dietz syndrome 6, 619656 (3), Autosomal dominant; Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 (3), Autosomal dominant
SMAD3	99.99 %	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	99.97 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMAD6	100 %	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMAD7	99.97 %	602932	{Colorectal cancer, susceptibility to, 3}, 612229 (3)
SMAD9	99.99 %	603295	Pulmonary hypertension, primary, 2, 615342 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SMARCA2	99.95 %	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant
SMARCA4	99.99 %	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant
SMARCAD1	99.86 %	612761	Basan syndrome, 129200 (3), Autosomal dominant; Huriez syndrome, 181600 (3), Autosomal dominant; Adermatoglyphia, 136000 (3), Autosomal dominant
SMARCAL1	99.97 %	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SMARCB1	99.99 %	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant
SMARCC1	99.92 %	601732	{Hydrocephalus, congenital, 5, susceptibility to}, 620241 (3), Autosomal dominant
SMARCC2	99.73 %	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
SMARCD1	99.81 %	601735	Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant
SMARCD2	99.99 %	601736	Specific granule deficiency 2, 617475 (3), Autosomal recessive
SMARCE1	99.87 %	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SMC1A	99.98 %	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant
SMC3	99.91 %	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMC5	99.47 %	609386	Atelis syndrome 2, 620185 (3), Autosomal recessive
SMCHD1	99.83 %	614982	Facioscapulohumeral muscular dystrophy 2, digenic, 158901 (3), Digenic dominant; Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant
SMG8	99.92 %	613175	Alzahrani-Kuwahara syndrome, 619268 (3), Autosomal recessive
SMG9	99.99 %	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive; Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995 (3), Autosomal recessive
SMIM1	100 %	615242	[Blood group, Vel system], 615264 (3), Autosomal recessive
SMN1	7.6 %	600354	Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive
SMN2	6.68 %	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
SMO	99.99 %	601500	Pallister-Hall-like syndrome, 241800 (3), Autosomal recessive; Basal cell carcinoma, somatic, 605462 (3); Curry-Jones syndrome, somatic mosaic, 601707 (3)
SMOC1	100 %	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMOC2	99.99 %	607223	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3), Autosomal recessive
SMPD1	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SMPD4	99.9 %	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive
SMPX	99.8 %	300226	Myopathy, distal, 7, adult-onset, X-linked, 301075 (3), X-linked recessive; Deafness, X-linked 4, 300066 (3), X-linked dominant
SMS	98.56 %	300105	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive
SNAP25	99.89 %	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SNAP29	99.85 %	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNAPC4	100 %	602777	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 (3), Autosomal recessive
SNCA	99.97 %	163890	Dementia, Lewy body, 127750 (3), Autosomal dominant; Parkinson disease 1, 168601 (3), Autosomal dominant; Parkinson disease 4, 605543 (3), Autosomal dominant
SNCB	99.99 %	602569	Dementia, Lewy body, 127750 (3), Autosomal dominant
SNF8	99.78 %	610904	Developmental and epileptic encephalopathy 115, 620783 (3), Autosomal recessive; Neurodevelopmental disorder plus optic atrophy, 620784 (3), Autosomal recessive
SNIP1	99.96 %	608241	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive
SNORA31	99.79 %	619378	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396 (3), Autosomal dominant
SNORD118	100 %	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
SNRNP200	99.6 %	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
SNRPB	99.96 %	182282	Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant
SNRPE	99.26 %	128260	Hypotrichosis 11, 615059 (3), Autosomal dominant
SNRPN	99.97 %	182279	*No OMIM phenotypes*
SNTA1	99.99 %	601017	Long QT syndrome 12, 612955 (3), Autosomal dominant
SNUPN	99.99 %	607902	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793 (3), Autosomal recessive
SNX10	99.96 %	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
SNX14	99.73 %	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
SOBP	99.99 %	613667	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
SOCS1	99.98 %	603597	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375 (3), Autosomal dominant
SOD1	99.97 %	147450	Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal dominant, Autosomal recessive
SOD2	99.98 %	147460	{Microvascular complications of diabetes 6}, 612634 (3)
SOD3	100 %	185490	[Superoxide dismutase, elevated extracellular] (3)
SOHLH1	100 %	610224	Ovarian dysgenesis 5, 617690 (3), Autosomal recessive; Spermatogenic failure 32, 618115 (3), Autosomal dominant
SON	99.95 %	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
SORD	85.52 %	182500	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912 (3), Autosomal recessive
SORT1	98.45 %	602458	[Low density lipoprotein cholesterol level QTL6], 613589 (3), Autosomal dominant
SOS1	99.68 %	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	99.39 %	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOST	100 %	605740	Sclerosteosis 1, 269500 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant
SOX10	100 %	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SOX11	100 %	600898	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 (3), Autosomal dominant
SOX17	100 %	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
SOX18	100 %	601618	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant
SOX2	100 %	184429	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant
SOX3	100 %	313430	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX4	99.36 %	184430	Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant
SOX5	99.96 %	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
SOX6	99.89 %	607257	Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant
SOX9	100 %	608160	Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant; Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant
SP110	99.99 %	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
SP6	100 %	608613	Amelogenesis imperfecta, type IK, 620104 (3), Autosomal dominant
SP7	100 %	606633	Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive
SPACA1	99.86 %	612739	?Spermatogenic failure 85, 620490 (3), Autosomal recessive
SPAG1	99.78 %	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
SPAG17	98.52 %	616554	?Spermatogenic failure 55, 619380 (3), Autosomal recessive
SPARC	99.94 %	182120	Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive
SPART	99.98 %	607111	Troyer syndrome, 275900 (3), Autosomal recessive
SPAST	99.77 %	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
SPATA16	99.96 %	609856	?Spermatogenic failure 6, 102530 (3), Autosomal recessive
SPATA5	99.82 %	613940	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive
SPATA5L1	99.91 %	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
SPATA7	99.8 %	609868	Leber congenital amaurosis 3, 604232 (3), Autosomal recessive; Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 (3), Autosomal recessive
SPECC1L	99.98 %	614140	Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant
SPEF2	99.93 %	610172	Spermatogenic failure 43, 618751 (3), Autosomal recessive
SPEG	99.99 %	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive
SPEN	99.98 %	613484	Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant
SPG11	99.89 %	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPG21	99.97 %	608181	Mast syndrome, 248900 (3), Autosomal recessive
SPG7	99.99 %	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal dominant, Autosomal recessive
SPI1	99.81 %	165170	Agammaglobulinemia 10, autosomal dominant, 619707 (3), Autosomal dominant
SPIDR	99.96 %	615384	Ovarian dysgenesis 9, 619665 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SPIN4	100 %	301113	?Lui-Jee-Baron syndrome, 301114 (3), X-linked
SPINK1	99.92 %	167790	Tropical calcific pancreatitis, 608189 (3), Autosomal dominant, Autosomal recessive; Pancreatitis, hereditary, 167800 (3), Autosomal dominant; {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 (3), Autosomal dominant, Autosomal recessive
SPINK2	100 %	605753	?Spermatogenic failure 29, 618091 (3), Autosomal recessive
SPINK5	99.91 %	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPINT2	99.9 %	605124	Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive
SPNS2	100 %	612584	?Deafness, autosomal recessive 115, 618457 (3), Autosomal recessive
SPOP	99.85 %	602650	Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant
SPPL2A	99.88 %	608238	Immunodeficiency 86, mycobacteriosis, 619549 (3), Autosomal recessive
SPR	99.99 %	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SPRED1	99.99 %	609291	Legius syndrome, 611431 (3), Autosomal dominant
SPRED2	99.99 %	609292	Noonan syndrome 14, 619745 (3), Autosomal recessive
SPRTN	99.98 %	616086	Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive
SPRY2	99.99 %	602466	{?IgA nephropathy, susceptibility to, 3}, 616818 (3), Autosomal dominant
SPRY4	100 %	607984	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant
SPTA1	99.08 %	182860	Spherocytosis, type 3, 270970 (3), Autosomal recessive; Elliptocytosis-2, 130600 (3), Autosomal dominant; Pyropoikilocytosis, 266140 (3), Autosomal recessive
SPTAN1	99.96 %	182810	Developmental delay with or without epilepsy, 620540 (3), Autosomal dominant; Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant; Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528 (3), Autosomal dominant
SPTB	100 %	182870	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 (3), Autosomal dominant, Autosomal recessive; Elliptocytosis-3, 617948 (3), Autosomal dominant, Autosomal recessive; Spherocytosis, type 2, 616649 (3), Autosomal dominant
SPTBN1	99.98 %	182790	Developmental delay, impaired speech, and behavioral abnormalities, 619475 (3), Autosomal dominant
SPTBN2	99.98 %	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SPTBN4	99.91 %	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
SPTLC1	99.74 %	605712	Amyotrophic lateral sclerosis 27, juvenile, 620285 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
SPTLC2	99.95 %	605713	Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3), Autosomal dominant
SPTSSA	99.88 %	613540	Spastic paraplegia 90A, autosomal dominant, 620416 (3), Autosomal dominant; ?Spastic paraplegia 90B, autosomal recessive, 620417 (3), Autosomal dominant
SQOR	99.99 %	617658	Sulfide:quinone oxidoreductase deficiency, 619221 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SQSTM1	100 %	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
SRC	99.99 %	190090	?Thrombocytopenia 6, 616937 (3), Autosomal dominant; Colon cancer, advanced, somatic, 114500 (3)
SRCAP	99.99 %	611421	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 (3), Autosomal dominant; Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRD5A2	100 %	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
SRD5A3	99.94 %	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
SREBF1	99.91 %	184756	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 (3), Autosomal dominant; Mucoepithelial dysplasia, hereditary, 158310 (3), Autosomal dominant
SRGAP1	99.68 %	606523	{Thyroid cancer, nonmedullary, 2}, 188470 (3), Somatic mutation, Autosomal dominant
SRP54	99.83 %	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
SRP68	99.99 %	604858	?Neutropenia, severe congenital, 10, autosomal recessive, 620534 (3), Autosomal recessive
SRP72	99.91 %	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
SRPX2	99.95 %	300642	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643 (3)
SRRM2	99.98 %	606032	Intellectual developmental disorder, autosomal dominant 72, 620439 (3), Autosomal dominant
SRSF1	99.99 %	600812	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489 (3), Autosomal dominant
SRY	51.98 %	480000	46XY sex reversal 1, 400044 (3), Y-linked; 46XX sex reversal 1, 400045 (4), X-linked dominant
SS18	99.98 %	600192	Sarcoma, synovial (1)
SSBP1	100 %	600439	Optic atrophy 13 with retinal and foveal abnormalities, 165510 (3), Autosomal dominant
SSR4	99.99 %	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
SSX1	99.97 %	312820	Spermatogenic failure, X-linked, 5, 301099 (3), X-linked
SSX2	21.85 %	300192	?Sarcoma, synovial, 300813 (3)
ST14	100 %	606797	Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive
ST3GAL3	99.98 %	606494	Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive
ST3GAL5	99.96 %	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAB1	99.98 %	608560	[Hyperferritinemia], 620729 (3), Autosomal recessive
STAC3	99.91 %	615521	Congenital myopathy 13, 255995 (3), Autosomal recessive
STAG1	99.88 %	604358	Intellectual developmental disorder, autosomal dominant 47, 617635 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
STAG2	99.08 %	300826	Holoprosencephaly 13, X-linked, 301043 (3), X-linked recessive, X-linked dominant; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked
STAG3	98.99 %	608489	Spermatogenic failure 61, 619672 (3), Autosomal recessive; Premature ovarian failure 8, 615723 (3), Autosomal recessive
STAMBP	99.95 %	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
STAR	99.97 %	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
STARD7	99.6 %	616712	Epilepsy, familial adult myoclonic, 2, 607876 (3), Autosomal dominant
STAT1	99.83 %	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive
STAT2	99.89 %	600556	Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive; Immunodeficiency 44, 616636 (3), Autosomal recessive
STAT3	99.97 %	102582	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STAT4	99.77 %	600558	Disabling pansclerotic morphea of childhood, 620443 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
STAT5B	99.5 %	604260	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3), Autosomal recessive; Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3), Autosomal dominant; Leukemia, acute promyelocytic, somatic, 102578 (3)
STAT6	99.87 %	601512	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections, 620532 (3), Autosomal dominant
STEAP3	100 %	609671	?Anemia, hypochromic microcytic, with iron overload 2, 615234 (3), Autosomal dominant
STEEP1	99.89 %	301012	?Intellectual developmental disorder, X-linked 107, 301013 (3), X-linked
STIL	99.11 %	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STIM1	99.99 %	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
STING1	99.87 %	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
STK11	100 %	602216	Melanoma, malignant, somatic, 155600 (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
STK33	99.93 %	607670	?Spermatogenic failure 93, 620849 (3)
STK36	99.98 %	607652	?Ciliary dyskinesia, primary, 46, 619436 (3), Autosomal recessive
STK4	99.91 %	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3), Autosomal recessive
STN1	99.88 %	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
STOX1	97.17 %	609397	Preeclampsia/eclampsia 4, 609404 (3), Autosomal dominant
STRA6	99.95 %	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
STRADA	99.98 %	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
STRC	45.5 %	606440	Deafness, autosomal recessive 16, 603720 (3), Autosomal recessive
STS	99.81 %	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
STT3A	99.99 %	601134	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive
STT3B	99.96 %	608605	Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
STUB1	99.99 %	607207	Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
STX11	100 %	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
STX16	100 %	603666	Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant
STX1B	99.97 %	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
STX3	100 %	600876	Retinal dystrophy and microvillus inclusion disease, 619446 (3), Autosomal recessive; Diarrhea 12, with microvillus atrophy, 619445 (3), Autosomal recessive
STX4	99.86 %	186591	?Deafness, autosomal recessive 123, 620745 (3), Autosomal recessive
STX5	99.95 %	603189	?Congenital disorder of glycosylation, type IIaa, 620454 (3), Autosomal recessive
STXBP1	99.99 %	602926	Developmental and epileptic encephalopathy 4, 612164 (3), Autosomal dominant, Autosomal recessive
STXBP2	100 %	601717	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3), Autosomal recessive
SUCLA2	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	99.64 %	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUFU	100 %	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal dominant, Autosomal recessive
SUGCT	99.35 %	609187	Glutaric aciduria III, 231690 (3), Autosomal recessive
SULT2B1	99.92 %	604125	Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive
SUMF1	99.95 %	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUMO1	98.45 %	601912	?Orofacial cleft 10, 613705 (3), Isolated cases
SUMO4	98.77 %	608829	{Diabetes mellitus, insulin-dependent, 5}, 600320 (3)
SUN5	100 %	613942	Spermatogenic failure 16, 617187 (3), Autosomal recessive
SUOX	100 %	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SUPT16H	99.97 %	605012	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 (3), Autosomal dominant
SURF1	100 %	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
SUZ12	98.58 %	606245	Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant
SV2A	99.65 %	185860	Developmental and epileptic encephalopathy 113, 620772 (3), Autosomal recessive
SVBP	99.04 %	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
SVIL	99.98 %	604126	Myofibrillar myopathy 10, 619040 (3), Autosomal recessive
SYCE1	99.99 %	611486	?Spermatogenic failure 15, 616950 (3), Autosomal recessive; ?Premature ovarian failure 12, 616947 (3), Autosomal recessive
SYCP2	99.67 %	604105	Spermatogenic failure 1, 258150 (3), Autosomal dominant
SYCP2L	97.48 %	616799	Premature ovarian failure 24, 620840 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SYCP3	98.98 %	604759	Pregnancy loss, recurrent, 4, 270960 (3), Autosomal dominant; Spermatogenic failure 4, 270960 (3), Autosomal dominant
SYK	99.96 %	600085	Immunodeficiency 82 with systemic inflammation, 619381 (3), Autosomal dominant
SYN1	99.98 %	313440	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491 (3), X-linked; Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked
SYN2	99.94 %	600755	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
SYNE1	99.95 %	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYNE2	99.95 %	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
SYNE4	99.99 %	615535	Deafness, autosomal recessive 76, 615540 (3), Autosomal recessive
SYNGAP1	99.96 %	603384	Intellectual developmental disorder, autosomal dominant 5, 612621 (3), Autosomal dominant
SYNJ1	99.91 %	604297	Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive
SYP	99.99 %	313475	Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive
SYT1	99.7 %	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
SYT14	99.94 %	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
SYT2	99.93 %	600104	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 (3), Autosomal dominant; Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461 (3), Autosomal recessive
SZT2	99.88 %	615463	Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive
TAB2	99.75 %	605101	Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant
TAC3	99.94 %	162330	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive
TACO1	100 %	612958	Mitochondrial complex IV deficiency, nuclear type 8, 619052 (3), Autosomal recessive
TACR3	99.99 %	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
TACSTD2	100 %	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
TAF1	99.84 %	313650	Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive
TAF13	95.71 %	600774	Intellectual developmental disorder, autosomal recessive 60, 617432 (3), Autosomal recessive
TAF15	99.91 %	601574	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TAF2	99.72 %	604912	Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive
TAF4	99.39 %	601796	Intellectual developmental disorder, autosomal dominant 73, 620450 (3), Autosomal dominant
TAF4B	99.97 %	601689	?Spermatogenic failure 13, 615841 (3), Autosomal recessive
TAF6	99.96 %	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
TAF8	99.91 %	609514	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 (3), Autosomal recessive
TAFAZZIN	99.98 %	300394	Barth syndrome, 302060 (3), X-linked recessive
TAL1	99.76 %	187040	Leukemia, T-cell acute lymphocytic, somatic, 613065 (3)
TAL2	99.98 %	186855	Leukemia, T-cell acute lymphocytic, somatic, 613065 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TALDO1	100 %	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
TAMM41	99.97 %	614948	Combined oxidative phosphorylation deficiency 56, 620139 (3), Autosomal recessive
TANC2	99.85 %	615047	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant
TANGO2	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TAOK1	99.77 %	610266	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 (3), Autosomal dominant
TAP1	99.97 %	170260	MHC class I deficiency 1, 604571 (3), Autosomal recessive
TAP2	99.94 %	170261	MHC class I deficiency 2, 620813 (3), Autosomal recessive
TAPBP	99.98 %	601962	?MHC class I deficiency 3, 620814 (3), Autosomal recessive
TAPT1	99.64 %	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive
TARDBP	100 %	605078	Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant; Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant
TARS1	99.92 %	187790	Trichothiodystrophy 7, nonphotosensitive, 618546 (3), Autosomal recessive
TARS2	99.77 %	612805	Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive
TAS2R16	100 %	604867	{Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial; [Beta-glycopyranoside tasting], 617956 (3), Autosomal dominant
TAS2R38	100 %	607751	[Phenylthiocarbamide tasting], 171200 (3), Autosomal dominant
TASP1	99.86 %	608270	Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive
TAT	99.99 %	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TBC1D20	100 %	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D23	98.7 %	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
TBC1D24	100 %	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
TBC1D2B	99.94 %	619152	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive
TBC1D32	99.75 %	615867	*No OMIM phenotypes*
TBC1D4	99.97 %	612465	{Diabetes mellitus, noninsulin-dependent, 5}, 616087 (3)
TBC1D7	99.9 %	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
TBC1D8B	99.31 %	301027	Nephrotic syndrome, type 20, 301028 (3), X-linked
TBCD	100 %	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
TBCE	99.91 %	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
TBCK	99.75 %	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TBK1	99.07 %	604834	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant; Autoinflammation with arthritis and vasculitis, 620880 (3), Autosomal recessive
TBL1X	99.96 %	300196	Hypothyroidism, congenital, nongoitrous, 8, 301033 (3), X-linked
TBL1XR1	99.92 %	608628	Intellectual developmental disorder, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
TBL1Y	51.79 %	400033	?Deafness, Y-linked 2, 400047 (3), Y-linked
TBP	99.98 %	600075	Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
TBR1	99.99 %	604616	Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant
TBX1	99.95 %	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TBX15	99.81 %	604127	Cousin syndrome, 260660 (3), Autosomal recessive
TBX18	99.5 %	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
TBX19	99.91 %	604614	Adrenocorticotropic hormone deficiency, 201400 (3), Autosomal recessive
TBX2	99.97 %	600747	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223 (3), Autosomal dominant
TBX20	99.99 %	606061	Atrial septal defect 4, 611363 (3)
TBX21	99.99 %	604895	Asthma and nasal polyps, 208550 (3), Autosomal recessive; ?Immunodeficiency 88, 619630 (3), Autosomal recessive; {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
TBX22	99.91 %	300307	Cleft palate with ankyloglossia, 303400 (3), X-linked; ?Abruzzo-Erickson syndrome, 302905 (3), X-linked
TBX3	100 %	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
TBX4	99.96 %	601719	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant; Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive
TBX5	99.98 %	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TBX6	99.99 %	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal dominant, Autosomal recessive
TBXA2R	98.85 %	188070	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009 (3), Autosomal dominant
TBXAS1	100 %	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive
TBXT	100 %	601397	Sacral agenesis with vertebral anomalies, 615709 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
TCAP	100 %	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive
TCEAL1	100 %	300237	Hijazi-Reis syndrome, 301094 (3), X-linked dominant
TCF12	99.97 %	600480	Craniosynostosis 3, 615314 (3), Autosomal dominant; Hypogonadotropic hypogonadism 26 with or without anosmia, 619718 (3), Autosomal dominant, Autosomal recessive
TCF20	100 %	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant
TCF3	100 %	147141	Agammaglobulinemia 8B, autosomal recessive, 619824 (3), Autosomal recessive; Agammaglobulinemia 8A, autosomal dominant, 616941 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TCF4	98.56 %	602272	Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant
TCF7L2	99.69 %	602228	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
TCHH	99.94 %	190370	?Uncombable hair syndrome 3, 617252 (3), Autosomal recessive
TCIRG1	99.99 %	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCL1A	100 %	186960	Leukemia/lymphoma, T-cell, 186960 (2)
TCL1B	100 %	603769	Leukemia/lymphoma, T-cell, 603769 (2)
TCN2	100 %	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TCOF1	99.99 %	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
TCTN1	99.92 %	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	99.99 %	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	99.92 %	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TDO2	99.89 %	191070	[?Hypertryptophanemia], 600627 (3), Autosomal recessive
TDP1	99.97 %	607198	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive
TDP2	99.97 %	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
TDRD7	99.87 %	611258	Cataract 36, 613887 (3), Autosomal recessive
TDRD9	99.95 %	617963	?Spermatogenic failure 30, 618110 (3), Autosomal recessive
TEAD1	99.97 %	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
TECPR2	99.96 %	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
TECR	99.99 %	610057	Intellectual developmental disorder, autosomal recessive 14, 614020 (3), Autosomal recessive
TECRL	99.66 %	617242	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 (3), Autosomal recessive
TECTA	99.99 %	602574	Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive
TEFM	99.88 %	616422	Combined oxidative phosphorylation deficiency 58, 620451 (3), Autosomal recessive
TEK	99.98 %	600221	Venous malformations, multiple cutaneous and mucosal, 600195 (3), Autosomal dominant; Glaucoma 3, primary congenital, E, 617272 (3), Autosomal dominant
TEKT3	100 %	612683	Spermatogenic failure 81, 620277 (3), Autosomal recessive
TELO2	99.99 %	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
TENM3	99.99 %	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
TENM4	99.99 %	610084	Essential tremor, hereditary, 5, 616736 (3), Autosomal dominant
TENT5A	99.97 %	611357	Osteogenesis imperfecta, type XVIII, 617952 (3), Autosomal recessive
TERB1	99.03 %	617332	Spermatogenic failure 60, 619646 (3), Autosomal recessive
TERB2	99.87 %	617131	?Spermatogenic failure 59, 619645 (3), Autosomal recessive
TERC	98.59 %	602322	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TERT	100 %	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal dominant, Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
TET2	99.99 %	612839	Myelodysplastic syndrome, somatic, 614286 (3); Immunodeficiency 75, 619126 (3), Autosomal recessive
TET3	99.95 %	613555	Beck-Fahrner syndrome, 618798 (3), Autosomal dominant, Autosomal recessive
TEX11	95.93 %	300311	Spermatogenic failure, X-linked 2, 309120 (3), X-linked recessive
TEX14	99.65 %	605792	Spermatogenic failure 23, 617707 (3), Autosomal recessive
TEX15	99.99 %	605795	Spermatogenic failure 25, 617960 (3), Autosomal recessive
TF	99.96 %	190000	Atransferrinemia, 209300 (3), Autosomal recessive
TFAM	99.24 %	600438	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive
TFAP2A	100 %	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TFAP2B	99.98 %	601601	Patent ductus arteriosus 2, 617035 (3), Autosomal dominant; Char syndrome, 169100 (3), Autosomal dominant
TFE3	99.87 %	314310	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 (3), X-linked; Renal cell carcinoma, papillary, 1, 300854 (3)
TFG	98.68 %	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
TFR2	99.97 %	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
TFRC	99.87 %	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
TG	99.99 %	188450	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive
TGDS	99.86 %	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
TGFB1	100 %	190180	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive
TGFB2	99.87 %	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
TGFB3	100 %	190230	Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant
TGFBI	99.89 %	601692	Corneal dystrophy, Avellino type, 607541 (3), Autosomal dominant; Corneal dystrophy, Reis-Bucklers type, 608470 (3), Autosomal dominant; Corneal dystrophy, Thiel-Behnke type, 602082 (3), Autosomal dominant; Corneal dystrophy, Groenouw type I, 121900 (3), Autosomal dominant; Corneal dystrophy, epithelial basement membrane, 121820 (3), Autosomal dominant; Corneal dystrophy, lattice type I, 122200 (3), Autosomal dominant; Corneal dystrophy, lattice type IIIA, 608471 (3), Autosomal dominant
TGFBR1	99.94 %	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	99.98 %	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
TGIF1	100 %	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
TGM1	99.82 %	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
TGM3	100 %	600238	?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TGM5	100 %	603805	Peeling skin syndrome 2, 609796 (3), Autosomal recessive
TGM6	99.99 %	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
TH	99.99 %	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THAP1	99.96 %	609520	Dystonia 6, torsion, 602629 (3), Autosomal dominant
THBD	100 %	188040	Thrombophilia 12 due to thrombomodulin defect, 614486 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
THBS2	99.99 %	188061	?Ehlers-Danlos syndrome, classic-like, 3, 620865 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3)
THG1L	99.99 %	618802	Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive
THOC1	99.96 %	606930	?Deafness, autosomal dominant 86, 620280 (3), Autosomal dominant
THOC2	99.11 %	300395	Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive
THOC6	99.94 %	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
THPO	100 %	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant; Thrombocytopenia 9, 620478 (3), Autosomal dominant; Amegakaryocytic thrombocytopenia, congenital, 2, 620481 (3), Autosomal recessive
THRA	100 %	190120	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant
THRB	99.82 %	190160	Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
THSD1	99.99 %	616821	?Aneurysm, intracranial berry, 12, 618734 (3), Autosomal dominant; Lymphatic malformation 13, 620244 (3), Autosomal recessive
THSD4	99.97 %	614476	Aortic aneurysm, familial thoracic 12, 619825 (3), Autosomal dominant
THUMPD1	99.79 %	616662	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989 (3), Autosomal recessive
TIA1	99.72 %	603518	Welander distal myopathy, 604454 (3), Autosomal dominant, Autosomal recessive; Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133 (3), Autosomal dominant
TIAM1	99.99 %	600687	Neurodevelopmental disorder with language delay and seizures, 619908 (3), Autosomal recessive
TICAM1	99.99 %	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal dominant, Autosomal recessive
TIE1	99.94 %	600222	Lymphatic malformation 11, 619401 (3), Autosomal dominant
TIMELESS	99.9 %	603887	?Advance sleep phase syndrome, familial, 4, 620015 (3), Autosomal dominant
TIMM22	100 %	607251	?Combined oxidative phosphorylation deficiency 43, 618851 (3), Autosomal recessive
TIMM50	99.99 %	607381	3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive
TIMM8A	100 %	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TIMMDC1	99.97 %	615534	Mitochondrial complex I deficiency, nuclear type 31, 618251 (3), Autosomal recessive
TIMP3	100 %	188826	Sorsby fundus dystrophy, 136900 (3), Autosomal dominant
TINF2	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TIRAP	100 %	606252	{Malaria, protection against}, 611162 (3); {Tuberculosis, protection against}, 607948 (3); {Bacteremia, protection against}, 614382 (3)
TJP2	99.99 %	607709	Hypercholanemia, familial 1, 607748 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TK2	99.96 %	188250	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 (3), Autosomal recessive; ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 (3), Autosomal recessive
TKFC	99.86 %	615844	Triokinase and FMN cyclase deficiency syndrome, 618805 (3), Autosomal recessive
TKT	97.39 %	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
TLCD3B	98.95 %	615175	Cone-rod dystrophy 22, 619531 (3), Autosomal recessive
TLE6	100 %	612399	Oocyte/zygote/embryo maturation arrest 15, 616814 (3), Autosomal recessive
TLK2	98.83 %	608439	Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant
TLL1	99.94 %	606742	Atrial septal defect 6, 613087 (3), Autosomal dominant
TLR1	100 %	601194	{Leprosy, susceptibility to, 5}, 613223 (3); {Leprosy, protection against}, 613223 (3)
TLR2	100 %	603028	{Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant; {Leprosy, susceptibility to}, 246300 (3), Autosomal dominant; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
TLR3	99.99 %	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal dominant, Autosomal recessive
TLR5	100 %	603031	{Melioidosis, susceptibility to}, 615557 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to, 1}, 601744 (3); {Systemic lupus erythematosus, resistance to}, 601744 (3); {Legionnaire disease, susceptibility to}, 608556 (3)
TLR7	99.98 %	300365	Immunodeficiency 74, COVID19-related, X-linked, 301051 (3), X-linked recessive; Systemic lupus erythematosus 17, 301080 (3), X-linked dominant
TLR8	99.98 %	300366	Immunodeficiency 98 with autoinflammation, X-linked, 301078 (3), X-linked, Somatic mosaicism
TM4SF20	99.99 %	615404	{Specific language impairment 5}, 615432 (3), Autosomal dominant
TMC1	99.87 %	606706	Deafness, autosomal dominant 36, 606705 (3), Autosomal dominant; Deafness, autosomal recessive 7, 600974 (3), Autosomal recessive
TMC6	100 %	605828	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400 (3), Autosomal recessive
TMC8	99.92 %	605829	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231 (3), Autosomal recessive
TMCO1	99.48 %	614123	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive
TMEM106B	99.92 %	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
TMEM107	100 %	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM126A	99.88 %	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
TMEM126B	99.77 %	615533	Mitochondrial complex I deficiency, nuclear type 29, 618250 (3), Autosomal recessive
TMEM127	99.99 %	613403	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
TMEM132E	100 %	616178	Deafness, autosomal recessive 99, 618481 (3), Autosomal recessive
TMEM138	100 %	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM147	100 %	613585	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075 (3), Autosomal recessive
TMEM151A	100 %	620108	Episodic kinesigenic dyskinesia 3, 620245 (3), Autosomal dominant
TMEM163	100 %	618978	Leukodystrophy, hypomyelinating, 25, 620243 (3), Autosomal dominant
TMEM165	99.97 %	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TMEM199	100 %	616815	Congenital disorder of glycosylation, type IIp, 616829 (3), Autosomal recessive
TMEM216	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM218	99.94 %	619285	Joubert syndrome 39, 619562 (3), Autosomal recessive
TMEM222	99.93 %	619469	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 (3), Autosomal recessive
TMEM231	88.88 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	99.3 %	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM240	99.99 %	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TMEM251	100 %	619332	Dysostosis multiplex, Ain-Naz type, 619345 (3), Autosomal recessive
TMEM260	99.9 %	617449	Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive
TMEM38B	99.95 %	611236	Osteogenesis imperfecta, type XIV, 615066 (3), Autosomal recessive
TMEM43	99.96 %	612048	Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Auditory neuropathy, autosomal dominant 3, 619832 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant
TMEM53	99.83 %	619722	Craniotubular dysplasia, Ikegawa type, 619727 (3), Autosomal recessive
TMEM63A	99.91 %	618685	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant
TMEM63C	99.99 %	619953	Spastic paraplegia 87, autosomal recessive, 619966 (3), Autosomal recessive
TMEM67	99.69 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TMEM70	99.99 %	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMEM94	99.96 %	618163	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive
TMEM98	100 %	615949	Nanophthalmos 4, 615972 (3), Autosomal dominant
TMIE	76.54 %	607237	Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive
TMLHE	77.23 %	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TMPRSS15	99.57 %	606635	Enterokinase deficiency, 226200 (3), Autosomal recessive
TMPRSS3	99.99 %	605511	Deafness, autosomal recessive 8/10, 601072 (3), Autosomal recessive
TMPRSS6	100 %	609862	Iron-refractory iron deficiency anemia, 206200 (3), Autosomal recessive
TMTC3	97.8 %	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
TMTC4	99.99 %	618203	?Deafness, autosomal recessive 122, 620714 (3), Autosomal recessive
TMX2	99.99 %	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
TNC	100 %	187380	Deafness, autosomal dominant 56, 615629 (3), Autosomal dominant
TNF	100 %	191160	{Migraine without aura, susceptibility to}, 157300 (3), Autosomal dominant; {Dementia, vascular, susceptibility to} (3); {Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Septic shock, susceptibility to} (3); {Malaria, cerebral, susceptibility to}, 611162 (3)
TNFAIP3	99.94 %	191163	Autoinflammatory syndrome, familial, Behcet-like 1, 616744 (3), Autosomal dominant
TNFRSF10B	99.98 %	603612	Squamous cell carcinoma, head and neck, 275355 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TNFRSF11A	100 %	603499	Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant
TNFRSF11B	100 %	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
TNFRSF13B	99.43 %	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal dominant, Autosomal recessive; Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	99.99 %	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
TNFRSF1A	100 %	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant
TNFRSF4	100 %	600315	?Immunodeficiency 16, 615593 (3), Autosomal recessive
TNFRSF9	99.99 %	602250	Immunodeficiency 109 with lymphoproliferation, 620282 (3), Autosomal recessive
TNFSF11	99.89 %	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
TNFSF4	100 %	603594	{Myocardial infarction, susceptibility to}, 608446 (3)
TNIK	99.93 %	610005	Intellectual developmental disorder, autosomal recessive 54, 617028 (3), Autosomal recessive
TNNC1	99.88 %	191040	Cardiomyopathy, dilated, 1Z, 611879 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 13, 613243 (3), Autosomal dominant
TNNC2	99.99 %	191039	Congenital myopathy 15, 620161 (3), Autosomal dominant
TNNI2	99.99 %	191043	Arthrogryposis, distal, type 2B1, 601680 (3), Autosomal dominant
TNNI3	100 %	191044	?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant; Cardiomyopathy, dilated, 1FF, 613286 (3)
TNNI3K	99.94 %	613932	Cardiac conduction disease with or without dilated cardiomyopathy, 616117 (3), Autosomal dominant
TNNT1	99.82 %	191041	Nemaline myopathy 5C, autosomal dominant, 620389 (3), Autosomal dominant; Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355 (3), Autosomal recessive; Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386 (3), Autosomal recessive
TNNT2	99.87 %	191045	Cardiomyopathy, dilated, 1D, 601494 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 2, 115195 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 3, 612422 (3), Autosomal dominant; Left ventricular noncompaction 6, 601494 (3), Autosomal dominant
TNNT3	100 %	600692	Arthrogryposis, distal, type 2B2, 618435 (3), Autosomal dominant
TNPO2	99.99 %	603002	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556 (3), Autosomal dominant
TNPO3	99.96 %	610032	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423 (3), Autosomal dominant
TNR	99.8 %	601995	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 (3), Autosomal recessive
TNRC6A	99.48 %	610739	?Epilepsy, familial adult myoclonic, 6, 618074 (3), Autosomal dominant
TNRC6B	99.99 %	610740	Global developmental delay with speech and behavioral abnormalities, 619243 (3), Autosomal dominant
TNXB	90.86 %	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
TOE1	99.96 %	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
TOGARAM1	99.93 %	617618	Joubert syndrome 37, 619185 (3), Autosomal recessive
TOM1	100 %	604700	?Immunodeficiency 85 and autoimmunity, 619510 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TOMM7	100 %	607980	Garg-Mishra progeroid syndrome, 620601 (3), Autosomal recessive
TONSL	100 %	604546	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive
TOP1	99.98 %	126420	DNA topoisomerase I, camptothecin-resistant (3)
TOP2A	99.71 %	126430	DNA topoisomerase II, resistance to inhibition of, by amsacrine (3)
TOP2B	99.72 %	126431	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296 (3), Autosomal dominant
TOP3A	99.91 %	601243	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive
TOPORS	99.99 %	609507	Retinitis pigmentosa 31, 609923 (3), Autosomal dominant
TOR1A	100 %	605204	{Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant
TOR1AIP1	98.45 %	614512	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 (3), Autosomal recessive
TP53	99.98 %	191170	{Basal cell carcinoma 7}, 614740 (3), Autosomal dominant; {Adrenocortical carcinoma, pediatric}, 202300 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Nasopharyngeal carcinoma, somatic, 607107 (3); {Osteosarcoma}, 259500 (3), Somatic mutation; {Choroid plexus papilloma}, 260500 (3), Autosomal dominant; {Colorectal cancer}, 114500 (3), Somatic mutation, Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Somatic mutation, Autosomal dominant; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant
TP53RK	100 %	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
TP63	99.97 %	603273	Premature ovarian failure 21, 620311 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant
TP73	100 %	601990	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive
TPCN2	99.98 %	612163	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267 (3)
TPH2	99.33 %	607478	{?Attention deficit-hyperactivity disorder, susceptibility to, 7}, 613003 (3); {Unipolar depression, susceptibility to}, 608516 (3)
TPI1	99.95 %	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPK1	99.96 %	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TPM1	99.92 %	191010	Left ventricular noncompaction 9, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant
TPM2	100 %	190990	Arthrogryposis, distal, type 2B4, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 1A, 108120 (3), Autosomal dominant; Congenital myopathy 23, 609285 (3), Autosomal dominant
TPM3	83.21 %	191030	Congenital myopathy 4A, autosomal dominant, 255310 (3), Autosomal dominant; Congenital myopathy 4B, autosomal recessive, 609284 (3), Autosomal recessive
TPM4	99.99 %	600317	Bleeding disorder, platelet-type, 25, 620486 (3), Autosomal dominant
TPMT	99.9 %	187680	{Thiopurines, poor metabolism of, 1}, 610460 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TPO	100 %	606765	Thyroid dyshormonogenesis 2A, 274500 (3), Autosomal recessive
TPP1	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TPP2	99.89 %	190470	Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive
TPR	99.25 %	189940	?Intellectual developmental disorder, autosomal recessive 79, 620393 (3), Autosomal recessive
TPRKB	81.09 %	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
TPRN	99.69 %	613354	Deafness, autosomal recessive 79, 613307 (3), Autosomal recessive
TRAC	100 %	186880	Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive
TRAF3	99.97 %	601896	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849 (3), Autosomal dominant
TRAF3IP1	99.96 %	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TRAF3IP2	100 %	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)
TRAF7	99.97 %	606692	Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant
TRAIP	99.97 %	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive
TRAK1	99.98 %	608112	Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive
TRAP1	100 %	606219	*No OMIM phenotypes*
TRAPPC10	84.42 %	602103	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027 (3), Autosomal recessive
TRAPPC11	99.93 %	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
TRAPPC12	99.96 %	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
TRAPPC14	99.99 %	618350	?Microcephaly 25, primary, autosomal recessive, 618351 (3), Autosomal recessive
TRAPPC2	99.41 %	300202	Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive
TRAPPC2L	100 %	610970	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive
TRAPPC4	100 %	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive
TRAPPC6B	99.74 %	610397	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive
TRAPPC9	99.98 %	611966	Intellectual developmental disorder, autosomal recessive 13, 613192 (3), Autosomal recessive
TRDN	99.8 %	603283	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 (3), Autosomal recessive
TREH	100 %	275360	Trehalase deficiency, 612119 (3), Autosomal recessive
TREM2	100 %	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal recessive
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
TRH	99.99 %	613879	Thyrotropin-releasing hormone deficiency, 275120 (1), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TRHR	100 %	188545	Hypothyroidism, congenital, nongoitrous, 7, 618573 (3), Autosomal recessive
TRIM2	99.98 %	614141	Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive
TRIM32	100 %	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRIM36	99.94 %	609317	?Anencephaly 1, 206500 (3), Autosomal recessive
TRIM37	98.19 %	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
TRIM44	99.99 %	612298	?Aniridia 3, 617142 (3), Autosomal dominant
TRIM71	100 %	618570	Hydrocephalus, congenital, 4, 618667 (3), Autosomal dominant
TRIM8	99.82 %	606125	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 (3), Autosomal dominant
TRIO	99.98 %	601893	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 (3), Autosomal dominant
TRIOBP	99.99 %	609761	Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive
TRIP11	99.9 %	604505	Odontochondrodysplasia 1, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive
TRIP12	99.62 %	604506	Intellectual developmental disorder, autosomal dominant 49, 617752 (3), Autosomal dominant
TRIP13	100 %	604507	Oocyte/zygote/embryo maturation arrest 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive
TRIP4	99.97 %	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive
TRIT1	99.25 %	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
TRMT1	100 %	611669	Intellectual developmental disorder, autosomal recessive 68, 618302 (3), Autosomal recessive
TRMT10A	99.9 %	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
TRMT10C	99.55 %	615423	Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive
TRMT5	99.99 %	611023	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539 (3), Autosomal recessive
TRMU	100 %	610230	{Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive
TRNT1	99.97 %	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
TRPA1	99.67 %	604775	?Episodic pain syndrome, familial, 1, 615040 (3), Autosomal dominant
TRPC3	99.97 %	602345	?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant
TRPC6	99.99 %	603652	Glomerulosclerosis, focal segmental, 2, 603965 (3), Autosomal dominant
TRPM1	99.96 %	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3), Autosomal recessive
TRPM3	99.89 %	608961	?Cataract 50 with or without glaucoma, 620253 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224 (3), Autosomal dominant
TRPM4	99.99 %	606936	Progressive familial heart block, type IB, 604559 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 6, 618531 (3), Autosomal dominant
TRPM6	99.93 %	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TRPM7	99.8 %	605692	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500 (3), Autosomal dominant
TRPS1	100 %	604386	Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant
TRPV3	99.9 %	607066	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant; Olmsted syndrome 1, 614594 (3), Autosomal dominant
TRPV4	100 %	605427	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); SED, Maroteaux type, 184095 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant
TRPV6	99.99 %	606680	Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive
TRRAP	99.79 %	603015	?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant
TSC1	99.99 %	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	99.98 %	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TSEN15	99.57 %	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
TSEN2	99.98 %	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
TSEN34	100 %	608754	?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive
TSEN54	100 %	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TSFM	100 %	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
TSGA10	98.49 %	607166	?Spermatogenic failure 26, 617961 (3), Autosomal recessive
TSHB	99.95 %	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
TSHR	100 %	603372	Hyperthyroidism, familial gestational, 603373 (3), Autosomal dominant; Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant; Thyroid adenoma, hyperfunctioning, somatic, 609152 (3); Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive; Thyroid carcinoma with thyrotoxicosis, somatic, 609152 (3)
TSHZ1	100 %	614427	Aural atresia, congenital, 607842 (3), Autosomal dominant
TSPAN12	99.89 %	613138	Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant
TSPAN7	99.97 %	300096	Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive
TSPEAR	100 %	612920	Tooth agenesis, selective, 10, 620173 (3), Autosomal recessive; ?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive; Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 (3), Autosomal recessive
TSPOAP1	99.95 %	610764	Dystonia 22, juvenile-onset, 620453 (3), Autosomal recessive; ?Dystonia 22, adult-onset, 620456 (3), Autosomal recessive
TSPYL1	100 %	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
TSR2	99.96 %	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TTBK2	99.93 %	611695	Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant
TTC12	99.97 %	610732	Ciliary dyskinesia, primary, 45, 618801 (3), Autosomal recessive
TTC19	99.99 %	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTC21A	99.98 %	611430	Spermatogenic failure 37, 618429 (3), Autosomal recessive
TTC21B	99.5 %	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
TTC26	99.97 %	617453	Biliary, renal, neurologic, and skeletal syndrome, 619534 (3), Autosomal recessive
TTC29	99.84 %	618735	Spermatogenic failure 42, 618745 (3), Autosomal recessive
TTC37	99.82 %	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TTC5	100 %	619014	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive
TTC7A	99.77 %	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
TTC8	99.67 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTI1	100 %	614425	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445 (3), Autosomal recessive
TTI2	99.94 %	614426	Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive
TTLL5	99.95 %	612268	Cone-rod dystrophy 19, 615860 (3), Autosomal recessive
TTN	99.15 %	188840	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Congenital myopathy 5 with cardiomyopathy, 611705 (3), Autosomal recessive; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Cardiomyopathy, dilated, 1G, 604145 (3), Autosomal dominant; Myopathy, myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant
TTPA	99.88 %	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TTR	100 %	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
TUB	100 %	601197	?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive
TUBA1A	99.97 %	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBA3D	99.94 %	617878	Keratoconus 9, 617928 (3), Autosomal dominant
TUBA4A	100 %	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
TUBA8	100 %	605742	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840 (3), Autosomal dominant
TUBB	100 %	191130	Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant
TUBB1	100 %	612901	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112 (3), Autosomal dominant
TUBB2A	87.26 %	615101	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
TUBB2B	87.69 %	612850	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TUBB3	100 %	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
TUBB4A	100 %	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TUBB4B	100 %	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant
TUBB6	99.98 %	615103	?Facial palsy, congenital, with ptosis and velopharyngeal dysfunction, 617732 (3), Autosomal dominant
TUBB8	99.83 %	616768	Oocyte/zygote/embryo maturation arrest 2, 616780 (3), Autosomal dominant, Autosomal recessive
TUBG1	99.95 %	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
TUBGCP2	96.54 %	617817	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive
TUBGCP4	99.8 %	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	100 %	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TUFM	100 %	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
TUFT1	99.51 %	600087	Woolly hair-skin fragility syndrome, 620415 (3), Autosomal recessive
TULP1	99.99 %	602280	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
TULP3	99.91 %	604730	Hepatorenocardiac degenerative fibrosis, 619902 (3), Autosomal recessive
TUSC3	99.96 %	601385	Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive
TWIST1	100 %	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant
TWIST2	100 %	607556	Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant; Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive
TWNK	100 %	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
TXN2	99.88 %	609063	?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive
TXNDC15	99.78 %	617778	Meckel syndrome 14, 619879 (3), Autosomal recessive
TXNL4A	99.99 %	611595	Burn-McKeown syndrome, 608572 (3), Autosomal recessive
TXNRD2	99.97 %	606448	?Glucocorticoid deficiency 5, 617825 (3), Autosomal recessive
TYK2	99.99 %	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive
TYMP	99.87 %	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
TYMS	99.83 %	188350	Dyskeratosis congenita, digenic, 620040 (3), Digenic dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TYR	100 %	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; Albinism, oculocutaneous, type IB, 606952 (3), Autosomal recessive; Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive
TYROBP	99.95 %	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
TYRP1	99.97 %	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive
U2AF2	99.98 %	191318	Developmental delay, dysmorphic facies, and brain anomalies, 620535 (3), Autosomal dominant
UBA1	99.93 %	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3)
UBA2	99.84 %	613295	ACCES syndrome, 619959 (3), Autosomal dominant
UBA5	99.95 %	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
UBAP1	99.51 %	609787	Spastic paraplegia 80, autosomal dominant, 618418 (3), Autosomal dominant
UBAP2L	99.45 %	616472	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494 (3), Autosomal dominant
UBE2A	99.77 %	312180	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive
UBE2T	99.89 %	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
UBE3A	99.23 %	601623	Angelman syndrome, 105830 (3), Autosomal dominant
UBE3B	99.99 %	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
UBE3C	99.97 %	614454	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270 (3), Autosomal recessive
UBE4A	99.99 %	603753	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639 (3), Autosomal recessive
UBIAD1	99.98 %	611632	Corneal dystrophy, Schnyder type, 121800 (3), Autosomal dominant
UBQLN2	100 %	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant
UBR1	99.93 %	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UBR7	99.96 %	613816	Li-Campeau syndrome, 619189 (3), Autosomal recessive
UBTF	99.99 %	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
UCHL1	99.99 %	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant; Spastic paraplegia 79A, autosomal dominant, 620221 (3), Autosomal dominant; Spastic paraplegia 79B, autosomal recessive, 615491 (3), Autosomal recessive
UCP2	99.95 %	601693	{Obesity, susceptibility to, BMIQ4}, 607447 (3)
UCP3	99.99 %	602044	{Obesity, severe, and type II diabetes}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive
UFC1	99.35 %	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
UFM1	99.25 %	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
UFSP2	99.74 %	611482	?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant; Developmental and epileptic encephalopathy 106, 620028 (3), Autosomal recessive
UGDH	99.77 %	603370	Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
UGP2	99.85 %	191760	Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive
UGT1A1	99.98 %	191740	Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; [Bilirubin, serum level of, QTL1], 601816 (3); Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal dominant, Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive
UGT2B17	88.99 %	601903	{Bone mineral density QTL 12, osteoporosis}, 612560 (3)
UMOD	99.98 %	191845	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000 (3), Autosomal dominant
UMPS	99.9 %	613891	Orotic aciduria, 258900 (3), Autosomal recessive
UNC119	100 %	604011	Cone-rod dystrophy 24, 620342 (3), Autosomal dominant; ?Immunodeficiency 13, 615518 (3), Autosomal dominant
UNC13D	100 %	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
UNC45A	100 %	611219	Osteootohepatoenteric syndrome, 619377 (3), Autosomal recessive
UNC45B	100 %	611220	?Cataract 43, 616279 (3), Autosomal dominant; Myofibrillar myopathy 11, 619178 (3), Autosomal recessive
UNC80	98.04 %	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
UNC93B1	99.75 %	608204	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3), Autosomal recessive
UNG	100 %	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
UPB1	100 %	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
UPF3B	99.26 %	300298	Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive
UQCC2	99.99 %	614461	Mitochondrial complex III deficiency, nuclear type 7, 615824 (3), Autosomal recessive
UQCC3	100 %	616097	?Mitochondrial complex III deficiency, nuclear type 9, 616111 (3), Autosomal recessive
UQCRB	99.51 %	191330	Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive
UQCRC1	99.99 %	191328	Parkinsonism with polyneuropathy, 619279 (3), Autosomal dominant
UQCRC2	99.49 %	191329	Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive
UQCRFS1	100 %	191327	Mitochondrial complex III deficiency, nuclear type 10, 618775 (3), Autosomal recessive
UQCRH	99.95 %	613844	?Mitochondrial complex III deficiency, nuclear type 11, 620137 (3), Autosomal recessive
UQCRQ	99.96 %	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
UROC1	99.99 %	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
UROD	99.32 %	613521	Porphyria, hepatoerythropoietic, 176100 (3), Autosomal dominant, Autosomal recessive; Porphyria cutanea tarda, 176100 (3), Autosomal dominant, Autosomal recessive
UROS	100 %	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
USB1	89.62 %	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
USF1	99.76 %	191523	{Hyperlipidemia, familial combined, susceptibility to}, 602491 (3)
USH1C	99.99 %	605242	Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive
USH1G	100 %	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
USH2A	99.88 %	608400	Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
USP18	93.05 %	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
USP26	100 %	300309	Spermatogenic failure, X-linked, 6, 301101 (3), X-linked
USP27X	99.99 %	300975	Intellectual developmental disorder, X-linked 105, 300984 (3), X-linked recessive
USP45	99.56 %	618439	?Leber congenital amaurosis 19, 618513 (3), Autosomal recessive
USP48	97.24 %	617445	Deafness, autosomal dominant 85, 620227 (3), Autosomal dominant
USP53	99.92 %	617431	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658 (3), Autosomal recessive
USP7	99.9 %	602519	Hao-Fountain syndrome, 616863 (3), Autosomal dominant
USP8	99.67 %	603158	Pituitary adenoma 4, ACTH-secreting, somatic, 219090 (3)
USP9X	99.84 %	300072	Intellectual developmental disorder, X-linked 99, 300919 (3), X-linked recessive; Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
USP9Y	51.65 %	400005	Spermatogenic failure, Y-linked, 2, 415000 (3), Y-linked
UVSSA	100 %	614632	UV-sensitive syndrome 3, 614640 (3), Autosomal recessive
VAC14	99.91 %	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
VAMP1	100 %	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VAMP2	100 %	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
VANGL1	98.4 %	610132	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant; Caudal regression syndrome, 600145 (3), Autosomal dominant
VANGL2	99.23 %	600533	Neural tube defects, 182940 (3), Autosomal dominant
VAPB	100 %	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant
VARS1	99.99 %	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive
VARS2	99.98 %	612802	Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive
VAX1	100 %	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
VCAN	99.99 %	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant
VCL	99.76 %	193065	Cardiomyopathy, dilated, 1W, 611407 (3); Cardiomyopathy, hypertrophic, 15, 613255 (3), Autosomal dominant
VCP	99.99 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant
VDR	99.86 %	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VEGFA	99.99 %	192240	{Microvascular complications of diabetes 1}, 603933 (3)
VEGFC	99.96 %	601528	Lymphatic malformation 4, 615907 (3), Autosomal dominant
VEZF1	99.96 %	606747	?Cardiomyopathy, dilated, 1OO, 620247 (3), Autosomal dominant
VHL	100 %	608537	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
VIM	100 %	193060	Cataract 30, pulverulent, 116300 (3), Autosomal dominant
VIPAS39	99.87 %	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VKORC1	100 %	608547	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3), Autosomal recessive; Warfarin resistance, 122700 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
VLDLR	99.99 %	192977	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive
VMA21	99.9 %	300913	Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive
VPS11	99.99 %	608549	?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
VPS13A	99.68 %	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
VPS13B	99.9 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS13C	99.65 %	608879	Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive
VPS13D	99.96 %	608877	Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive
VPS16	100 %	608550	Dystonia 30, 619291 (3), Autosomal dominant
VPS33A	95.13 %	610034	Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive
VPS33B	99.95 %	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
VPS35	99.83 %	601501	{Parkinson disease 17}, 614203 (3), Autosomal dominant
VPS35L	99.07 %	618981	Ritscher-Schinzel syndrome 3, 619135 (3), Autosomal recessive
VPS37A	99.96 %	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
VPS41	99.92 %	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
VPS45	93.94 %	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
VPS4A	99.98 %	609982	CIMDAG syndrome, 619273 (3), Autosomal dominant
VPS50	98.35 %	616465	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 (3), Autosomal recessive
VPS51	93 %	615738	Pontocerebellar hypoplasia, type 13, 618606 (3), Autosomal recessive
VPS53	100 %	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
VRK1	99.98 %	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 (3), Autosomal recessive
VSX1	99.93 %	605020	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3), Autosomal dominant; Keratoconus 1, 148300 (3), Autosomal dominant
VSX2	99.99 %	142993	Microphthalmia, isolated 2, 610093 (3), Autosomal recessive; Microphthalmia with coloboma 3, 610092 (3), Autosomal recessive
VWA1	99.99 %	611901	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216 (3), Autosomal recessive
VWA3B	98.82 %	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
VWA8	99.8 %	617509	?Retinitis pigmentosa 97, 620422 (3), Autosomal dominant
VWF	98.1 %	613160	von Willebrand disease, type 1, 193400 (3), Autosomal dominant; von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3), Autosomal dominant, Autosomal recessive; von Willebrand disease, type 3, 277480 (3), Autosomal recessive
WAC	99.76 %	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
WARS1	99.97 %	191050	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317 (3), Autosomal recessive
WARS2	97.72 %	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
WAS	99.9 %	300392	Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive
WASF1	99.86 %	605035	Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant
WASHC4	99.84 %	615748	Intellectual developmental disorder, autosomal recessive 43, 615817 (3), Autosomal recessive
WASHC5	99.98 %	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WBP11	99.88 %	618083	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227 (3), Autosomal dominant
WBP2	99.99 %	606962	Deafness, autosomal recessive 107, 617639 (3), Autosomal recessive
WBP4	99.95 %	604981	Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852 (3), Autosomal recessive
WDFY3	99.93 %	617485	?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant
WDPCP	99.87 %	613580	Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR1	99.99 %	604734	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive
WDR11	99.91 %	606417	Intellectual developmental disorder, autosomal recessive 78, 620237 (3), Autosomal recessive; Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant
WDR19	99.8 %	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR26	96.29 %	617424	Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant
WDR35	99.92 %	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WDR36	99.46 %	609669	Glaucoma 1, open angle, G, 609887 (3)
WDR37	99.94 %	618586	Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant
WDR4	99.95 %	605924	Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive
WDR45	99.99 %	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR45B	100 %	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
WDR62	99.96 %	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
WDR72	96.51 %	613214	Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive
WDR73	99.92 %	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WDR81	100 %	614218	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
WEE2	99.98 %	614084	Oocyte/zygote/embryo maturation arrest 5, 617996 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
WFS1	99.99 %	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
WHRN	99.96 %	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
WIPF1	99.87 %	602357	Wiskott-Aldrich syndrome 2, 614493 (3), Autosomal recessive
WIPI2	99.99 %	609225	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 (3), Autosomal recessive
WLS	99.6 %	611514	Zaki syndrome, 619648 (3), Autosomal recessive
WNK1	99.98 %	605232	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant
WNK3	99.82 %	300358	Prieto syndrome, 309610 (3), X-linked recessive
WNK4	99.98 %	601844	Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant
WNT1	99.97 %	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3), Autosomal dominant; Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
WNT10A	100 %	606268	Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal dominant, Autosomal recessive; Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980 (3), Autosomal recessive
WNT10B	99.97 %	601906	Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (3), Autosomal recessive
WNT2B	99.88 %	601968	Diarrhea 9, 618168 (3), Autosomal recessive
WNT3	99.99 %	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
WNT4	99.94 %	603490	?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant
WNT5A	100 %	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
WNT7A	99.99 %	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive
WNT7B	100 %	601967	*No OMIM phenotypes*
WRAP53	100 %	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
WRN	99.83 %	604611	Werner syndrome, 277700 (3), Autosomal recessive
WT1	99.99 %	607102	Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant
WWC1	96.2 %	610533	[Memory, enhanced, QTL], 615602 (3)
WWOX	100 %	605131	Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XBP1	99.99 %	194355	{Major affective disorder-7, susceptibility to}, 612371 (3)
XDH	99.97 %	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
XIAP	99.36 %	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive
XIST	99.22 %	314670	X-inactivation, familial skewed, 300087 (3), X-linked
XK	99.98 %	314850	McLeod syndrome, 300842 (3), X-linked
XPA	99.68 %	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
XPC	99.98 %	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
XPNPEP2	99.95 %	300145	{Angioedema induced by ACE inhibitors, susceptibility to}, 300909 (3)
XPNPEP3	99.99 %	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XPR1	99.13 %	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant
XRCC1	99.96 %	194360	?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive
XRCC2	99.99 %	600375	Spermatogenic failure 50, 619145 (3), Autosomal recessive; ?Premature ovarian failure 17, 619146 (3), Autosomal recessive; ?Fanconi anemia, complementation group U, 617247 (3), Autosomal recessive
XRCC3	100 %	600675	{Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Melanoma, cutaneous malignant, 6}, 613972 (3)
XRCC4	99.89 %	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
XYLT1	99.98 %	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
XYLT2	99.97 %	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
YAP1	99.87 %	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433 (3), Autosomal dominant
YARS1	99.29 %	603623	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 (3), Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant
YARS2	99.93 %	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
YEATS2	99.96 %	613373	?Epilepsy, myoclonic, familial adult, 4, 615127 (3), Autosomal dominant
YIF1B	99.8 %	619109	Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive
YIPF5	99.99 %	611483	Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive
YME1L1	99.87 %	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
YRDC	99.85 %	612276	Galloway-Mowat syndrome 10, 619609 (3), Autosomal recessive
YWHAG	99.99 %	605356	Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant
YY1	100 %	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
YY1AP1	99.98 %	607860	Grange syndrome, 602531 (3), Autosomal recessive
ZAP70	99.95 %	176947	Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive
ZBTB11	99.97 %	618181	Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive
ZBTB16	100 %	176797	Leukemia, acute promyelocytic, PL2F/RARA type (3)
ZBTB18	99.41 %	608433	Intellectual developmental disorder, autosomal dominant 22, 612337 (3), Autosomal dominant
ZBTB20	100 %	606025	Primrose syndrome, 259050 (3), Autosomal dominant
ZBTB24	99.99 %	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
ZBTB42	100 %	613915	?Lethal congenital contracture syndrome 6, 616248 (3), Autosomal recessive
ZBTB7A	100 %	605878	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769 (3), Autosomal dominant
ZC3H14	99.96 %	613279	Intellectual developmental disorder, autosomal recessive 56, 617125 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ZC4H2	99.98 %	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
ZCCHC8	99.95 %	616381	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674 (3), Autosomal dominant
ZDHHC9	99.61 %	300646	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked
ZEB1	100 %	189909	Corneal dystrophy, posterior polymorphous, 3, 609141 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 6, 613270 (3), Autosomal dominant
ZEB2	99.97 %	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
ZFAT	99.96 %	610931	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3)
ZFHX2	99.99 %	617828	?Marsili syndrome, 147430 (3), Autosomal dominant
ZFHX3	99.96 %	104155	Prostate cancer, somatic, 176807 (3); {Atrial fibrillation 8, susceptibility to}, 613055 (3), Autosomal dominant; Spinocerebellar ataxia 4, 600223 (3), Autosomal dominant
ZFHX4	99.99 %	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
ZFP36L2	100 %	612053	Oocyte/zygote/embryo maturation arrest 13, 620154 (3), Autosomal recessive
ZFP57	100 %	612192	Diabetes mellitus, transient neonatal 1, 601410 (3), Autosomal dominant, Autosomal recessive
ZFPM2	100 %	603693	Diaphragmatic hernia 3, 610187 (3); 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
ZFX	94.74 %	314980	Intellectual developmental disorder, X-linked syndromic 37, 301118 (3), X-linked
ZFYVE19	99.99 %	619635	Cholestasis, progressive familial intrahepatic, 9, 619849 (3), Autosomal recessive
ZFYVE26	100 %	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
ZIC1	99.99 %	600470	?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant
ZIC2	100 %	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
ZIC3	99.9 %	300265	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZMIZ1	99.92 %	607159	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant
ZMPSTE24	98.7 %	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive
ZMYM2	99.88 %	602221	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant
ZMYM3	99.98 %	300061	Intellectual developmental disorder, X-linked 112, 301111 (3), X-linked recessive
ZMYND10	99.99 %	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
ZMYND11	99.99 %	608668	Intellectual developmental disorder, autosomal dominant 30, 616083 (3), Autosomal dominant
ZMYND15	100 %	614312	?Spermatogenic failure 14, 615842 (3), Autosomal recessive
ZNF141	97.64 %	194648	?Polydactyly, postaxial, type A6, 615226 (3), Autosomal recessive
ZNF142	100 %	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive
ZNF148	99.85 %	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
ZNF292	99.91 %	616213	Intellectual developmental disorder, autosomal dominant 64, 619188 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ZNF335	100 %	610827	Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive
ZNF341	100 %	618269	Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282 (3), Autosomal recessive
ZNF365	99.97 %	607818	{Nephrolithiasis, uric acid, susceptibility to}, 605990 (3)
ZNF407	99.98 %	615894	SIMHA syndrome, 619557 (3), Autosomal recessive
ZNF408	99.99 %	616454	Retinitis pigmentosa 72, 616469 (3), Autosomal recessive; ?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant
ZNF423	98.94 %	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive
ZNF462	99.99 %	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
ZNF469	100 %	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive
ZNF513	99.97 %	613598	?Retinitis pigmentosa 58, 613617 (3), Autosomal recessive
ZNF526	100 %	614387	Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive
ZNF644	99.37 %	614159	Myopia 21, autosomal dominant, 614167 (3), Autosomal dominant
ZNF668	99.99 %	617103	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194 (3), Autosomal recessive
ZNF687	100 %	610568	Paget disease of bone 6, 616833 (3), Autosomal dominant
ZNF699	100 %	609571	DEGCAGS syndrome, 619488 (3), Autosomal recessive
ZNF711	99.43 %	314990	Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked
ZNF750	100 %	610226	?Seborrhea-like dermatitis with psoriasiform elements, 610227 (3)
ZNFX1	99.99 %	618931	Immunodeficiency 91 and hyperinflammation, 619644 (3), Autosomal recessive
ZNHIT3	62.89 %	604500	PEHO syndrome, 260565 (3), Autosomal recessive
ZP1	99.94 %	195000	Oocyte/zygote/embryo maturation arrest 1, 615774 (3), Autosomal recessive
ZP2	99.79 %	182888	Oocyte/zygote/embryo maturation arrest 6, 618353 (3), Autosomal recessive
ZP3	93.11 %	182889	Oocyte/zygote/embryo maturation arrest 3, 617712 (3), Autosomal dominant
ZPBP	99.08 %	608498	?Spermatogenic failure 66, 619799 (3), Autosomal recessive
ZPR1	99.97 %	603901	?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321 (3), Autosomal recessive
ZSCAN10	100 %	618365	Otofacial neurodevelopmental syndrome, 620910 (3), Autosomal dominant
ZSWIM6	98.94 %	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant
ZSWIM7	99.99 %	614535	Spermatogenic failure 71, 619831 (3), Autosomal recessive; ?Ovarian dysgenesis 10, 619834 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.