Whole Exome Sequencing
Gene package Metabolic disorders, version 5.1, 31-1-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	63	100	97	90
ABAT	GABA-transaminase deficiency, 613163	137150	100	100	100	96
ABCA1	HDL deficiency, familial, 1, 604091 Tangier disease, 205400	600046	92	100	100	97
ABCB11	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847	603201	67	100	100	96
ABCB4	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803	171060	69	100	99	92
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	59	100	98	87
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	86	84	78	75
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	108	100	100	100
ABCG2	[Junior blood group system], 614490 [Uric acid concentration, serum, QTL1], 138900	603756	69	100	99	92

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ABCG5	Sitosterolemia, 210250	605459	80	100	100	99
ABCG8	{Gallbladder disease 4}, 611465 Sitosterolemia, 210250	605460	149	100	98	95
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	61	100	100	94
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	74	100	100	98
ACACA	Acetyl-CoA carboxylase deficiency, 613933	200350	73	100	100	95
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	159	100	100	100
ACAD9	Mitochondrial complex I deficiency, nuclear type 20, 611126	611103	95	100	100	100
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	607008	66	100	100	96
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	606885	121	100	100	100
ACADSB	2-methylbutyrylglycinuria, 610006	600301	67	100	100	94
ACADVL	VLCAD deficiency, 201475	609575	114	100	100	98
ACAT1	Alpha-methylacetoacetic aciduria, 203750	607809	71	100	100	93
ACAT2	?ACAT2 deficiency, 614055	100678	92	100	100	98
ACBD5	No OMIM phenotype	616618	85	100	99	89
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	146	100	98	94
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	103	100	100	99
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	149	100	100	100
ACY1	Aminoacylase 1 deficiency, 609924	104620	109	100	100	100
ADA	Adenosine deaminase deficiency, partial, 102700, Somatic mosaicism Severe combined immunodeficiency due to ADA deficiency, 102700, Somatic mosaicism	608958	85	100	100	96
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	600293	111	98	95	93
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	53	100	93	80
ADSL	Adenylosuccinase deficiency, 103050	608222	89	100	100	96
AGA	Aspartylglucosaminuria, 208400	613228	73	100	100	96
AGK	Cataract 38, 614691 Sengers syndrome, 212350	610345	57	100	100	93
AGL	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400	610860	72	100	100	95
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	142	100	100	96
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121	603051	61	100	99	90
AGXT	Hyperoxaluria, primary, type 1, 259900	604285	133	100	100	100
AGXT2	[Beta-aminoisobutyric acid, urinary excretion of], 210100	612471	71	100	99	92
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	151	96	96	96

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AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness 5, 300614	300169	55	100	96	82
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	124	100	100	100
AK2	Reticular dysgenesis, 267500	103020	68	100	100	97
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555	604741	64	100	100	97
ALAD	{Lead poisoning, susceptibility to}, 612740 Porphyria, acute hepatic, 612740	125270	104	100	100	100
ALAS2	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, 300752	301300	60	100	98	90
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	80	100	100	98
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	88	100	100	97
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	64	100	99	93
ALDH4A1	Hyperprolinemia, type II, 239510	606811	118	100	100	99
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	66	100	97	91
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	603178	98	100	100	98
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	69	100	100	91
ALDOA	Glycogen storage disease XII, 611881	103850	144	100	100	100
ALDOB	Fructose intolerance, hereditary, 229600	612724	102	100	100	100
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	61	91	79	73
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	73	100	100	99
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	158	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	50	100	97	83
ALG14	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227	612866	102	100	100	97
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	81	100	100	99
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	95	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	73	100	100	96
ALG8	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874	608103	62	100	100	94

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ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	64	100	100	94
ALOX12B	Ichthyosis, congenital 2, 242100	603741	113	100	100	99
ALPL	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300	171760	130	100	100	100
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	96	100	100	98
AMN	Megaloblastic anemia-1, Norwegian type, 261100	605799	89	100	93	83
AMPD1	Myopathy due to myoadenylate deaminase deficiency, 615511	102770	67	100	99	95
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686	102771	142	100	100	100
AMT	Glycine encephalopathy, 605899	238310	130	100	100	100
APOA1	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 618463 Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463	107680	130	100	100	100
APOB	Hypercholesterolemia, familial, 2, 144010 Hypobetalipoproteinemia, 615558	107730	159	100	100	100
APOC2	Hyperlipoproteinemia, type Ib, 207750	608083	91	100	100	100
APOE	Alzheimer disease-2, 104310 {Coronary artery disease, severe, susceptibility to}, 617347 Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 {?Macular degeneration, age-related}, 603075 Sea-blue histiocyte disease, 269600	107741	102	100	100	95
APRT	Adenine phosphoribosyltransferase deficiency, 614723	102600	81	100	100	100
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	76	100	99	92
ARG1	Argininemia, 207800	608313	75	100	100	98
ARSA	Metachromatic leukodystrophy, 250100	607574	136	100	100	100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	611542	73	100	100	98
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	67	100	100	93
ASL	Argininosuccinic aciduria, 207900	608310	117	100	100	99
ASNS	Asparagine synthetase deficiency, 615574	108370	73	100	100	92

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ASPA	Canavan disease, 271900	608034	55	100	99	91
ASS1	Citrullinemia, 215700	603470	115	100	98	90
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	62	100	100	96
ATP5F1E	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	606153	88	100	100	100
ATP6AP1	Immunodeficiency 47, 300972	300197	76	100	99	95
ATP6V0A2	Cutis laxa, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	81	100	100	97
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	49	100	98	84
ATP7B	Wilson disease, 277900	606882	99	100	100	99
ATP8B1	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600	602397	70	100	100	95
ATPAF2	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	73	100	100	100
AUH	3-methylglutaconic aciduria, type I, 250950	600529	94	100	100	98
B3GALNT1	[Blood group, P1PK system, P(k) phenotype], 111400 [Blood group, globoside system], 615021	603094	89	100	100	100
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	59	100	100	97
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	64	79	75	72
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600	606374	125	100	100	100
B3GLCT	Peters-plus syndrome, 261540	610308	67	100	99	85
B4GALNT1	Spastic paraplegia 26, 609195	601873	111	100	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	95	100	100	100
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	604327	126	100	100	98
BAAT	Hypercholanemia, familial, 607748	602938	73	100	100	87
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	160	100	100	100
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	64	100	99	94
BCO1	?Hypercarotenemia and vitamin A deficiency, 115300	605748	74	100	100	97
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	170	100	100	100

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BHMT	No OMIM phenotype	602888	58	100	95	86
BLVRA	Hyperbiliverdinemia, 614156	109750	78	100	100	97
BMP2	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877	112261	123	100	100	99
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299	613183	69	100	100	91
BPGM	Erythrocytosis, familial, 8, 222800	613896	71	100	100	99
BTD	Biotinidase deficiency, 253260	609019	89	100	100	100
C1GALT1C1	Tn polyagglutination syndrome, somatic, 300622	300611	47	100	100	95
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	175	100	100	100
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	121	100	100	100
CANT1	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719	613165	128	100	100	100
CAT	Acatalasemia, 614097	115500	70	100	100	93
CBLIF	Intrinsic factor deficiency, 261000	609342	No coverage data			
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	127	100	100	100
CCDC115	Congenital disorder of glycosylation, type IIo, 616828	613734	65	87	82	79
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646	606475	95	100	100	100
CEL	Maturity-onset diabetes of the young, type VIII, 609812	114840	151	94	90	87
CERKL	Retinitis pigmentosa 26, 608380	608381	73	100	99	94
CERS3	Ichthyosis, congenital 9, 615023	615276	65	100	100	94
CETP	[High density lipoprotein cholesterol level QTL 10], 143470 Hyperalphalipoproteinemia, 143470	118470	95	100	100	100
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	111	100	100	100
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1, 601776	608429	133	100	98	97
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	603799	129	100	100	100
CHST6	Macular corneal dystrophy, 217800	605294	246	100	100	100
CHSY1	Temtamy preaxial brachydactyly syndrome, 605282	608183	79	98	95	92
CIC	Mental retardation 45, 617600	612082	134	100	100	99
CLDN16	Hypomagnesemia 3, renal, 248250	603959	73	100	100	97
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	149	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	111	100	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	70	100	100	100

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CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	120	100	100	99
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	129	100	100	100
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	114	100	100	100
CLPS	No OMIM phenotype	120105	114	100	100	100
CLPX	?Protoporphyria, erythropoietic, 2, 618015	615611	55	100	98	85
CNDP1	No OMIM phenotype	609064	72	100	100	96
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	161	100	100	99
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	96	100	100	98
COG2	?Congenital disorder of glycosylation, type IIq, 617395	606974	65	100	100	93
COG4	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150	606976	81	100	100	99
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	66	100	99	91
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	65	100	99	90
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	83	100	100	98
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	125	100	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	66	100	100	94
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	129	100	100	100
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	139	100	100	99
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	65	100	98	92
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	102	100	100	100
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	63	100	100	96
CPOX	Coproporphyria, 121300 Harderoporphyria, 121300	612732	82	100	100	98
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371	608307	61	100	100	96
CPT1A	CPT deficiency, hepatic, type IA, 255120	600528	109	100	100	98

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CPT2	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212	600650	106	100	100	99
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	81	100	99	93
CRTC1	Mucoepidermoid salivary gland carcinoma	607536	146	100	100	99
CTH	Cystathioninuria, 219500 Homocysteine, total plasma, elevated	607657	76	100	100	98
CTNS	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750	606272	118	100	100	100
CTSA	Galactosialidosis, 256540	613111	124	100	100	100
CTSC	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650	602365	75	100	100	97
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	137	100	100	100
CTSK	Pycnodysostosis, 265800	601105	65	100	100	98
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	82	100	100	96
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	127	100	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	131	100	100	100
CYP27B1	Vitamin D-dependent rickets, type I, 264700	609506	122	100	100	100
CYP7A1	No OMIM phenotype	118455	69	100	100	98
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, 270800	603711	67	100	100	98
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	140	100	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	63	100	100	97
DBH	Orthostatic hypotension 1, due to DBH deficiency, 223360	609312	136	100	100	100
DBT	Maple syrup urine disease, type II, 248600	248610	115	100	100	99
DCXR	[Pentosuria], 260800	608347	165	100	100	100
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	84	100	99	92
DDHD1	Spastic paraplegia 28, 609340	614603	107	100	98	92
DDOST	?Congenital disorder of glycosylation, type Ir, 614507	602202	92	100	100	100

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DGAT1	?Diarrhea 7, protein-losing enteropathy type, 615863	604900	116	96	91	85
DGKE	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008	601440	106	100	100	98
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions 4, 617070	601465	73	100	100	92
DHCR24	Desmosterolosis, 602398	606418	119	100	100	100
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	112	100	100	100
DHDDS	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861	608172	69	100	100	99
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	76	100	100	98
DHODH	Miller syndrome, 263750	126064	87	100	100	96
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	85	100	100	98
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	74	100	98	90
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	77	100	100	98
DLST	Paragangliomas 7, 618475	126063	81	100	100	100
DMGDH	Dimethylglycine dehydrogenase deficiency, 605850	605849	75	100	100	99
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	69	100	95	88
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	75	100	100	96
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	217	100	100	100
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708	603850	70	100	99	94
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, 604121 Neuropathy, hereditary sensory, type IE, 614116	126375	93	100	100	97
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	136	100	100	100
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	76	100	100	100
DPEP1	No OMIM phenotype	179780	124	100	100	100
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	79	94	89	85
DPM2	Congenital disorder of glycosylation, type Iu, 615042	603564	76	100	100	100
DPM3	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937	605951	227	100	100	100
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	66	100	99	95

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DPYS	Dihydropyrimidinuria, 222748	613326	75	100	100	98
EBP	Chondrodysplasia punctata dominant, 302960 MEND syndrome, 300960	300205	93	100	100	100
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277	602292	79	100	100	100
EGF	Hypomagnesemia 4, renal, 611718	131530	74	100	100	97
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	71	100	100	98
ENO3	?Glycogen storage disease XIII, 612932	131370	148	100	100	100
EOGT	Adams-Oliver syndrome 4, 615297	614789	60	100	100	94
EPG5	Vici syndrome, 242840	615068	69	100	100	97
EPHX1	?Hypercholanemia, familial, 607748	132810	128	100	100	99
EPHX2	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890	132811	88	100	100	95
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	86	89	87	85
ETFA	Glutaric acidemia IIA, 231680	608053	60	100	100	96
ETFB	Glutaric acidemia IIB, 231680	130410	106	100	100	100
ETFDH	Glutaric acidemia IIC, 231680	231675	85	100	100	99
ETHE1	Ethylmalonic encephalopathy, 602473	608451	113	100	100	98
EXT1	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700	608177	70	100	100	96
EXT2	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	98	100	100	97
FA2H	Spastic paraplegia 35, 612319	611026	83	100	99	91
FAH	Tyrosinemia, type I, 276700	613871	99	100	100	100
FASTKD2	?Mitochondrial complex IV deficiency, 220110	612322	74	100	100	96
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700	611570	119	100	100	100
FECH	Protoporphyria, erythropoietic, 1, 177000	612386	64	100	100	96

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FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579	176943	74	100	100	95
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	82	99	94	87
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	143	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	85	100	100	100
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100	610595	136	100	100	100
FMO3	Trimethylaminuria, 602079	136132	69	100	100	95
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	111	100	100	100
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19, 618241	613622	91	100	100	100
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	96	99	96	93
FUCA1	Fucosidosis, 230000	612280	83	100	100	98
FUT2	[Bombay phenotype, digenic], 616754 {Norwalk virus infection, resistance to} {Vitamin B12 plasma level QTL1}, 612542	182100	182	100	100	100
FXYD2	Hypomagnesemia 2, renal, 154020	601814	101	100	100	100
G6PC	Glycogen storage disease Ia, 232200	613742	113	100	100	100

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G6PC3	Dursun syndrome, 612541 Neutropenia, severe congenital 4, 612541	611045	103	100	100	100
G6PD	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162	305900	96	100	100	100
GAA	Glycogen storage disease II, 232300	606800	156	100	100	100
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	86	100	100	99
GALC	Krabbe disease, 245200	606890	54	100	99	89
GALE	Galactose epimerase deficiency, 230350	606953	116	100	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	133	100	100	99
GALNS	Mucopolysaccharidosis IVA, 253000	612222	81	100	99	95
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900	601756	69	100	100	97
GALT	Galactosemia, 230400	606999	151	100	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	85	100	99	95
GANAB	Polycystic kidney disease 3, 600666	104160	85	100	99	96
GART	No OMIM phenotype	138440	55	100	96	89
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	65	100	100	98
GBA	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600	606463	172	100	100	100
GBA2	Spastic paraplegia 46, 614409	609471	137	100	100	100
GBE1	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	607839	67	100	99	92
GCDH	Glutaricaciduria, type I, 231670	608801	112	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	52	100	100	93
GCK	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851	138079	127	100	100	100
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446	606857	88	100	100	97

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GCSH	?Glycine encephalopathy, 605899	238330	93	100	89	62
GFM1	Combined oxidative phosphorylation deficiency 1, 609060	606639	72	100	100	96
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, 610542	138292	59	100	100	92
GGT1	?Glutathioninuria, 231950	612346	54	77	63	57
GK	Glycerol kinase deficiency, 307030	300474	46	99	88	66
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	101	100	100	100
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	105	100	100	98
GLDC	Glycine encephalopathy, 605899	238300	67	100	98	91
GLRA1	Hyperekplexia 1, 149400	138491	85	100	100	96
GLRX5	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859	609588	68	100	91	81
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	99	100	97	91
GLUL	Glutamine deficiency, congenital, 610015	138290	92	100	100	100
GLYCTK	D-glyceric aciduria, 220120	610516	136	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	94	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	114	100	100	100
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352	615320	189	100	100	100
GNE	Nonaka myopathy, 605820 Sialuria, 269921	603824	93	100	100	99
GNMT	Glycine N-methyltransferase deficiency, 606664	606628	105	100	100	100
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	78	100	100	96
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	63	100	99	95
GNPTG	Mucolipidosis III gamma, 252605	607838	153	100	99	94
GNS	Mucopolysaccharidosis type IIID, 252940	607664	68	100	100	98
GPD1	Hypertriglyceridemia, transient infantile, 614480	138420	80	100	100	100
GPHN	Molybdenum cofactor deficiency C, 615501	603930	68	100	100	98
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470	172400	130	100	100	100
GPX1	Hemolytic anemia due to glutathione peroxidase deficiency, 614164	138320	95	100	100	100

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GRHPR	Hyperoxaluria, primary, type II, 260000	604296	96	100	100	100
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	89	100	100	99
GSTZ1	[Maleylacetoacetate isomerase deficiency], 617596	603758	95	100	100	100
GUSB	Mucopolysaccharidosis VII, 253220	611499	106	100	100	97
GYG1	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199	603942	60	100	100	96
GYS1	Glycogen storage disease 0, muscle, 611556	138570	116	100	100	100
GYS2	Glycogen storage disease 0, liver, 240600	138571	67	100	100	98
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	77	100	100	100
HADHA	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015	600890	99	100	100	97
HADHB	Trifunctional protein deficiency, 609015	143450	66	100	100	97
HAL	[Histidinemia], 235800	609457	89	100	100	97
HAMP	Hemochromatosis, type 2B, 613313	606464	161	100	100	100
HCFC1	Mental retardation 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	79	100	97	92
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	86	100	100	98
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	119	100	100	97
HFE	{Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200 [Transferrin serum level QTL2], 614193	613609	97	100	100	100
HGD	Alkaptonuria, 203500	607474	63	100	100	96
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	70	94	94	92
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	610690	51	100	94	75

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HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	102	100	100	99
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	98	100	100	99
HMBS	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000	609806	88	100	100	100
HMGCL	HMG-CoA lyase deficiency, 246450	613898	94	100	100	100
HMGCS2	HMG-CoA synthase-2 deficiency, 605911	600234	78	100	100	99
HMOX1	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	141250	112	100	100	99
HOGA1	Hyperoxaluria, primary, type III, 613616	613597	122	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	107	100	100	99
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	43	100	99	81
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	103	100	100	97
HS6ST1	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880	604846	119	100	100	97
HSD11B1	Cortisone reductase deficiency 2, 614662	600713	68	100	100	99
HSD11B2	Apparent mineralocorticoid excess, 218030	614232	147	92	86	83
HSD17B10	HSD10 mitochondrial disease, 300438	300256	76	100	100	100
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	62	100	98	93
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810	613890	143	100	100	100
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765	607764	115	100	100	100
HYAL1	?Mucopolysaccharidosis type IX, 601492	607071	103	100	100	100
IBA57	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, 616451	615316	124	100	100	97
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	105	100	100	100
IDH3B	Retinitis pigmentosa 46, 612572	604526	117	100	100	100
IDS	Mucopolysaccharidosis II, 309900	300823	74	100	99	95
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	138	100	97	91
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type, 614078	614010	146	100	100	97

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IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	99	100	96	91
INPPL1	Opsismodysplasia, 258480	600829	111	100	100	99
ISCU	Myopathy with lactic acidosis, hereditary, 255125	611911	75	100	100	100
ITPA	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850	147520	122	100	100	100
IVD	Isovaleric acidemia, 243500	607036	107	100	100	98
KHK	[Fructosuria], 229800	614058	119	100	100	100
KYNU	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661	605197	73	100	100	97
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	74	100	100	97
LAMP2	Danon disease, 300257	309060	44	100	95	77
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840	603590	97	100	100	99
LBR	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471	600024	68	100	100	96
LCAT	Fish-eye disease, 136120 Norum disease, 245900	606967	130	100	100	97
LCT	Lactase deficiency, congenital, 223000	603202	114	100	100	100
LDHA	Glycogen storage disease XI, 612933	150000	62	100	99	88
LDLR	Hypercholesterolemia, familial, 1, 143890 LDL cholesterol level QTL2, 143890	606945	209	100	100	100
LFNG	Spondylocostal dysostosis 3, 609813	602576	129	86	84	83
LIPA	Cholesteryl ester storage disease, 278000 Wolman disease, 278000	613497	68	100	100	96
LIPC	{Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797	151670	82	100	100	98
LIPI	No OMIM phenotype	609252	60	100	99	92
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380	612625	65	100	98	85
LPIN1	Myoglobinuria, acute recurrent, 268200	605518	78	100	100	97
LPIN2	Majeed syndrome, 609628	605519	77	100	100	98

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LPL	Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11], 238600 Lipoprotein lipase deficiency, 238600	609708	110	100	100	99
LRP2	Donnai-Barrow syndrome, 222448	600073	69	100	100	96
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	60	100	99	91
LTC4S	Leukotriene C4 synthase deficiency, 614037	246530	80	100	85	74
LYST	Chediak-Higashi syndrome, 214500	606897	72	100	99	95
MAN1B1	Mental retardation 15, 614202	604346	122	100	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	115	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	85	100	100	95
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	49	100	99	92
MAT1A	Hypermethioninemia, persistent, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, 250850	610550	107	100	100	100
MAT2A	No OMIM phenotype	601468	83	100	100	96
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	74	100	100	94
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	64	100	100	96
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120	608419	77	100	100	95
MCOLN1	Mucolipidosis IV, 252650	605248	129	100	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	68	100	100	95
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	104	100	100	100
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	65	100	96	92
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	78	100	100	98
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110	607568	82	100	100	98
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	133	100	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	63	100	100	90
MMUT	Methylmalonic aciduria, mut(0) type, 251000	609058	75	100	100	95
MOCOS	Xanthinuria, type II, 603592	613274	87	100	100	95
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	105	100	100	99
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	67	100	100	96
MOCS3	No OMIM phenotype	609277	176	100	100	100

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MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	130	100	100	100
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	87	100	100	99
MPI	Congenital disorder of glycosylation, type Ib, 602579	154550	154	100	100	99
MPV17	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	137960	108	100	100	100
MRPS16	Combined oxidative phosphorylation deficiency 2, 610498	609204	120	100	100	100
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117	605810	75	100	100	96
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834	607545	95	100	100	98
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	172460	76	100	100	96
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	99	100	100	99
MTM1	Myotubular myopathy, 310400	300415	46	100	95	75
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	80	100	100	97
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	87	100	100	98
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	67	100	100	97
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	100	100	100	100
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	118	100	100	100
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	112	100	97	93
NAGS	N-acetylglutamate synthase deficiency, 237310	608300	110	100	100	100
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	65	100	99	94
NAT8L	?N-acetylaspartate deficiency, 614063	610647	73	94	90	84
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12, 301020	300078	110	100	100	100

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NDUFA11	Mitochondrial complex I deficiency, nuclear type 14, 618236	612638	117	100	100	100
NDUFA2	?Mitochondrial complex I deficiency, nuclear type 13, 618235	602137	104	100	100	100
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11, 618234	606934	63	100	100	95
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10, 618233	609653	83	100	92	78
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18, 618240	612911	130	100	100	100
NDUFAF4	Mitochondrial complex I deficiency, nuclear type 15, 618237	611776	95	100	100	100
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16, 618238	612360	70	100	100	96
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5, 618226	157655	69	100	100	94
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6, 618228	602985	79	100	100	99
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8, 618230	603846	112	100	100	100
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1, 252010	602694	83	100	100	99
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9, 618232	603848	70	100	100	100
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3, 618224	601825	130	100	100	98
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2, 618222	602141	140	100	100	100
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4, 618225	161015	129	100	100	100
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7, 618229	600532	48	100	95	80
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	155	100	100	100
NFU1	Multiple mitochondrial dysfunctions syndrome 1, 605711	608100	66	100	100	95
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	144	100	100	100
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	87	100	100	99
NPC2	Niemann-pick disease, type C2, 607625	601015	95	100	100	100
NPRL2	Epilepsy, familial focal, with variable foci 2, 617116	607072	119	100	100	100
NR5A2	No OMIM phenotype	604453	81	100	100	98
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	68	100	100	93
NT5C	No OMIM phenotype	191720	93	100	81	72
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency, 266120	606224	61	100	99	89
NT5E	Calcification of joints and arteries, 211800	129190	90	100	100	98
NUBPL	Mitochondrial complex I deficiency, nuclear type 21, 618242	613621	60	100	100	97
NUS1	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation 55, with seizures, 617831	610463	54	100	98	78
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	64	100	96	85

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	43	100	95	80
OGDH	Alpha-ketoglutarate dehydrogenase deficiency, 203740	613022	131	100	100	99
OPA1	Behr syndrome, 210000 {Glaucoma, normal tension, susceptibility to}, 606657 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250	605290	59	100	98	89
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	130	100	100	100
OPLAH	5-oxoprolinase deficiency, 260005	614243	132	100	100	99
OTC	Ornithine transcarbamylase deficiency, 311250	300461	58	100	98	85
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	601424	74	100	100	96
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	68	100	99	95
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	81	100	100	99
PC	Pyruvate carboxylase deficiency, 266150	608786	141	100	100	100
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070	126090	87	100	100	99
PCCA	Propionicacidemia, 606054	232000	68	100	100	95
PCCB	Propionicacidemia, 606054	232050	72	100	98	95
PCK1	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680	614168	114	100	100	100
PCK2	PEPCK deficiency, 261650	614095	121	100	100	100
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	49	100	94	84
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	65	100	100	95
PDHX	Lacticacidemia due to PDX1 deficiency, 245349	608769	79	100	100	97
PDK1	No OMIM phenotype	602524	74	100	100	96
PDK2	No OMIM phenotype	602525	117	100	100	100
PDK3	?Charcot-Marie-Tooth disease dominant, 6, 300905	300906	52	100	98	82
PDK4	No OMIM phenotype	602527	57	100	100	93
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	87	100	100	100
PDP2	No OMIM phenotype	615499	126	100	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	60	100	93	85
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	75	100	97	87
PDXK	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511	179020	98	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PEPD	Prolidase deficiency, 170100	613230	100	100	100	98
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	62	100	99	94
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	100	100	100	97
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	137	100	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	67	100	100	94
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	69	100	100	99
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887	601791	142	100	100	100
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	134	100	96	94
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	67	100	100	98
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	63	100	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	118	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	57	100	100	95
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	111	100	100	100
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	107	100	99	95
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	57	100	100	95
PFKM	Glycogen storage disease VII, 232800	610681	92	100	100	99
PGAM2	Glycogen storage disease X, 261670	612931	156	100	100	100
PGAP1	Mental retardation 42, 615802	611655	65	100	99	90
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	147	100	100	100
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	115	100	100	100
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	47	100	100	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PGM1	Congenital disorder of glycosylation, type It, 614921	171900	72	100	100	99
PGM3	Immunodeficiency 23, 615816	172100	88	100	100	98
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	138	100	100	100
PHKA1	Muscle glycogenosis, 300559	311870	47	100	97	82
PHKA2	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000	300798	56	100	99	90
PHKB	Phosphorylase kinase deficiency of liver and muscle, 261750	172490	76	100	100	98
PHKG1	No OMIM phenotype	172470	123	100	100	97
PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027	172471	123	100	100	100
PHYH	Refsum disease, 266500	602026	121	100	100	94
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	67	100	100	97
PIGC	Glycosylphosphatidylinositol biosynthesis defect 16, 617816	601730	149	100	100	100
PIGG	Mental retardation 53, 616917	616918	100	100	100	98
PIGL	CHIME syndrome, 280000	605947	98	100	100	100
PIGM	Glycosylphosphatidylinositol deficiency, 610293	610273	85	100	100	100
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	63	100	99	89
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	126	100	100	100
PIGQ	Epileptic encephalopathy, early infantile, 77, 618548	605754	157	100	100	100
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	138	100	100	100
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	96	100	100	100
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11, 616025	610275	85	100	100	100
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	44	100	100	94
PIK3R1	?Agammaglobulinemia 7, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	80	100	100	97
PIK3R5	Ataxia-oculomotor apraxia 3, 615217	611317	107	100	100	100
PKLR	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200	609712	156	100	100	100
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	118	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	60	100	99	94
PLIN1	Lipodystrophy, familial partial, type 4, 613877	170290	96	100	100	99
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400	153454	100	100	100	99
PLOD2	Bruck syndrome 2, 609220	601865	58	100	99	90
PLOD3	Lysyl hydroxylase 3 deficiency, 612394	603066	100	100	100	99
PLPBP	Epilepsy, early-onset, vitamin B6-dependent, 617290	604436	53	100	97	84
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	72	100	100	97
PMPCA	Spinocerebellar ataxia 2, 213200	613036	133	100	100	100
PNLIP	?Pancreatic lipase deficiency, 614338	246600	60	100	100	95
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	70	100	100	97
PNPLA2	Neutral lipid storage disease with myopathy, 610717	609059	114	100	100	100
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	123	100	100	99
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	69	100	100	94
POFUT1	Dowling-Degos disease 2, 615327	607491	114	100	100	100
POGLUT1	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle 21, 617232	615618	54	100	98	91
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	114	100	100	99
POLG2	Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 Progressive external ophthalmoplegia with mitochondrial DNA deletions 4, 610131	604983	140	100	100	95
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090	614258	84	100	100	98
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	76	100	99	93
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123	606822	93	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135	614828	154	100	100	100
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094	615247	95	100	100	100
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	103	100	100	100
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	87	100	100	99
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	124015	170	100	100	100
PPM1K	?Maple syrup urine disease, mild variant, 615135	611065	77	100	100	98
PPOX	Porphyria variegata, 176200	600923	106	100	100	100
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	84	100	100	100
PRKAG2	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200	602743	123	100	100	97
PRKCSH	Polycystic liver disease 1, 174050	177060	142	100	100	98
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	98	100	95	86
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	50	100	99	90
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	121	100	100	97
PSAT1	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	610936	64	100	100	97
PSPH	Phosphoserine phosphatase deficiency, 614023	172480	51	100	96	84
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	86	100	100	95
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	88	100	99	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PYCR1	Cutis laxa, type IIB, 612940 Cutis laxa, type IIIB, 614438	179035	93	100	100	100
PYGL	Glycogen storage disease VI, 232700	613741	84	100	100	96
PYGM	McArdle disease, 232600	608455	112	100	100	100
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	94	100	100	97
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	66	100	99	93
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895	610924	135	100	100	99
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	128	100	100	100
RBSN	No OMIM phenotype	609511	110	100	100	97
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	67	100	99	93
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	109	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	62	100	98	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	323	100	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	106	100	100	99
RPIA	Ribose 5-phosphate isomerase deficiency, 608611	180430	77	100	100	98
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions 5, 613077	604712	90	100	100	100
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	90	100	100	94
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	64	100	99	88
SAR1B	Chylomicron retention disease, 246700	607690	79	100	100	97
SARDH	[Sarcosinemia], 268900	604455	104	100	100	99
SC5D	Lathosterolosis, 607330	602286	88	100	100	100
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	602257	89	100	100	98
SCN3A	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938	182391	84	100	100	97
SCO1	Mitochondrial complex IV deficiency, 220110	603644	99	100	100	99
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	125	100	100	100
SCP2	?Leukoencephalopathy with dystonia and motor neuropathy, 613724	184755	54	100	100	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	134	100	97	91
SDHAF1	Mitochondrial complex II deficiency, 252011	612848	82	100	100	100
SDHB	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300	185470	149	100	100	100
SDHC	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373	602413	170	100	100	100
SDHD	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300	602690	164	100	100	100
SEC23B	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100	610512	74	100	100	97
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	78	100	100	96
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	65	100	99	86
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	103	100	94	90
SHMT1	No OMIM phenotype	182144	95	100	100	100
SHMT2	No OMIM phenotype	138450	135	100	100	99
SI	Sucrase-isomaltase deficiency, congenital, 222900	609845	60	100	99	92
SLC12A3	Gitelman syndrome, 263800	600968	113	100	100	100
SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095	600682	83	100	100	99
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	86	100	100	97
SLC19A1	No OMIM phenotype	600424	167	100	99	97
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270	603941	68	100	100	99
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	73	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	93	100	100	98
SLC1A3	Episodic ataxia, type 6, 612656	600111	99	100	100	100
SLC22A5	Carnitine deficiency, systemic primary, 212140	603377	118	100	100	100
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 ?Myasthenic syndrome, congenital, 23, presynaptic, 618197	190315	102	100	100	100
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	88	100	100	97
SLC25A13	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814	603859	98	100	99	93
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	132	100	100	98
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	105	100	100	100
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138	613698	66	100	100	97
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	122	100	100	100
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773	600370	105	100	100	100
SLC25A32	?Exercise intolerance, riboflavin-responsive, 616839	610815	58	100	97	83
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950	610819	109	100	100	100
SLC25A4	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions 2, 609283	103220	112	100	100	100
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	127	100	100	100
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi-Bickel syndrome, 227810	138160	63	100	100	96
SLC30A10	Hypermanganesemia with dystonia 1, 613280	611146	148	100	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	68	100	98	86
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	66	100	100	97
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	73	100	99	92
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	146	100	100	100
SLC35D1	Schneckenbecken dysplasia, 269250	610804	57	100	99	85

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SLC36A2	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608331	102	100	100	100
SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	96	100	100	95
SLC39A14	Hypermanganesemia with dystonia 2, 617013 ?Hyperostosis cranalis interna, 144755	608736	92	100	100	97
SLC39A4	Acrodermatitis enteropathica, 201100	607059	125	100	100	100
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721	608732	83	100	99	95
SLC3A1	Cystinuria, 220100	104614	93	100	100	97
SLC40A1	Hemochromatosis, type 4, 606069	604653	63	100	100	97
SLC46A1	Folate malabsorption, hereditary, 229050	611672	121	100	100	98
SLC52A1	Riboflavin deficiency, 615026	607883	203	100	100	100
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	173	100	100	100
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500	613350	109	100	100	100
SLC5A1	Glucose/galactose malabsorption, 606824	182380	104	100	100	96
SLC5A2	Renal glucosuria, 233100	182381	113	100	100	99
SLC6A19	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608893	115	100	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	103	100	98	95
SLC7A7	Lysinuric protein intolerance, 222700	603593	79	100	100	97
SLC7A9	Cystinuria, 220100	604144	100	100	100	99
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic, 237450	604843	61	100	99	90
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic, 237450	605495	66	100	99	92
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	131	100	100	99
SOD1	Amyotrophic lateral sclerosis 1, 105400	147450	74	100	100	100
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	102	100	100	100
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA, 162400	605712	62	100	99	94
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640	605713	81	100	100	96
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	111	100	100	96
SSR3	No OMIM phenotype	606213	63	94	77	57
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	76	100	100	99

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ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation 12, 611090	606494	79	100	100	96
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	55	99	92	84
STAR	Lipoid adrenal hyperplasia, 201710	600617	118	100	100	100
STS	Ichthyosis, 308100	300747	90	97	97	95
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	63	100	100	98
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	90	100	100	97
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	56	100	97	86
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	88	100	100	100
SUGCT	Glutaric aciduria III, 231690	609187	73	100	100	93
SUMF1	Multiple sulfatase deficiency, 272200	607939	88	100	100	99
SUOX	Sulfite oxidase deficiency, 272300	606887	136	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	88	94	89	87
TACO1	Mitochondrial complex IV deficiency, 220110	612958	82	100	100	100
TALDO1	Transaldolase deficiency, 606003	602063	136	100	100	100
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	616830	96	100	100	99
TAT	Tyrosinemia, type II, 276600	613018	65	100	100	98
TAZ	Barth syndrome, 302060	300394	97	100	98	90
TBXAS1	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158	274180	74	100	99	93
TCIRG1	Osteopetrosis 1, 259700	604592	117	100	100	97
TCN1	No OMIM phenotype	189905	66	100	99	95
TCN2	Transcobalamin II deficiency, 275350	613441	118	100	100	100
TDO2	[?Hypertryptophanemia], 600627	191070	49	100	99	88
TECR	Mental retardation 14, 614020	610057	140	100	100	100
TFR2	Hemochromatosis, type 3, 604250	604720	101	100	100	97
TH	Segawa syndrome, recessive, 605407	191290	93	100	99	94
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	138	100	100	100
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 617069	188250	66	100	100	95
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	108	100	100	100
TMEM199	Congenital disorder of glycosylation, type IIp, 616829	616815	85	100	100	100

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TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	93	100	100	95
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	123	100	98	96
TPMT	{Thiopurines, poor metabolism of, 1}, 610460	187680	75	100	100	89
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	97	100	100	100
TRAPPC11	Muscular dystrophy, limb-girdle 18, 615356	614138	60	100	97	90
TREH	Trehalase deficiency, 612119	275360	111	100	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	234	100	100	100
TRIM37	Mulibrey nanism, 253250	605073	61	100	100	94
TRMU	{Deafness, modifier of}, 580000 Liver failure, transient infantile, 613070	610230	97	100	100	98
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	75	100	100	96
TSFM	Combined oxidative phosphorylation deficiency 3, 610505	604723	77	100	100	100
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	53	100	92	77
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	61	100	98	91
TUFM	Combined oxidative phosphorylation deficiency 4, 610678	602389	158	100	100	100
TUSC3	Mental retardation 7, 611093	601385	72	100	100	95
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	145	100	100	100
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	131222	102	100	100	100
TYMS	No OMIM phenotype	188350	76	100	100	97
TYR	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 Waardenburg syndrome/albinism, digenic, 103470	606933	83	100	100	98
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	73	100	100	98

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UGT1A1	[Bilirubin, serum level of, QTL1], 601816 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 [Gilbert syndrome], 143500 Hyperbilirubinemia, familial transient neonatal, 237900	191740	130	100	100	100
UMOD	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860	191845	95	100	100	97
UMPS	Orotic aciduria, 258900	613891	74	100	100	98
UPB1	Beta-ureidopropionase deficiency, 613161	606673	96	100	100	100
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158	191330	75	100	100	100
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	123	100	100	100
UROC1	?Urocanase deficiency, 276880	613012	112	100	100	99
UROD	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100	613521	87	100	100	97
UROS	Porphyria, congenital erythropoietic, 263700	606938	60	100	100	97
USF1	{Hyperlipidemia, familial combined, susceptibility to}, 602491	191523	113	100	100	100
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700	608547	113	100	100	99
VPS13B	Cohen syndrome, 216550	607817	75	100	99	96
XDH	Xanthinuria, type I, 278300	607633	81	100	100	96
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	103	100	97	93
XYLT2	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822	608125	131	100	100	100
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	610957	127	100	100	99

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x

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