Whole Exome Sequencing
Gene package Metabolic disorders, version 4, 18-2-2019

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

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
AASS	Hyperlysinemia, 238700 Saccharopinuria, 268700	605113	56	100	96	86
ABAT	GABA-transaminase deficiency, 613163	137150	95	100	99	94
ABCA1	{Coronary artery disease in familial hypercholesterolemia, protection against}, 143890 HDL deficiency, type 2, 604091 Tangier disease, 205400	600046	81	100	99	94
ABCB11	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847	603201	56	100	99	90
ABCB4	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803	171060	57	100	98	85
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	53	100	98	81
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	75	85	76	72
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	98	100	100	98
ABCG2	[Junior blood group system], 614490 [Uric acid concentration, serum, QTL1], 138900	603756	59	100	98	87
ABCG5	Sitosterolemia, 210250	605459	70	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
ABCG8	{Gallbladder disease 4}, 611465 Sitosterolemia, 210250	605460	138	100	97	95
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	57	100	100	91
ABHD5	Chanarin-Dorfman syndrome, 275630	604780	62	100	100	95
ACACA	Acetyl-CoA carboxylase deficiency, 613933	200350	64	100	99	89
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	163	100	100	100
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	88	100	100	99
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	607008	58	100	99	90
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	606885	118	100	100	100
ACADSB	2-methylbutyrylglycinuria, 610006	600301	59	100	99	86
ACADVL	VLCAD deficiency, 201475	609575	109	100	100	97
ACAT1	Alpha-methylacetoacetic aciduria, 203750	607809	62	100	98	89
ACAT2	?ACAT2 deficiency, 614055	100678	82	100	100	94
ACBD5	No OMIM phenotype	616618	76	100	98	82
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	138	100	97	94
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	95	100	100	98
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	144	100	100	99
ACY1	Aminoacylase 1 deficiency, 609924	104620	99	100	100	100
ADA	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700	608958	79	100	100	95
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	600293	109	99	96	93
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	44	100	91	70
ADSL	Adenylosuccinase deficiency, 103050	608222	78	100	99	92
AGA	Aspartylglucosaminuria, 208400	613228	62	100	99	92
AGK	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350	610345	49	100	98	88
AGL	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400	610860	59	100	99	90
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594	603100	128	100	100	99
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121	603051	56	100	97	82
AGXT	Hyperoxaluria, primary, type 1, 259900	604285	124	100	100	100
AGXT2	[Beta-aminoisobutyric acid, urinary excretion of], 210100	612471	63	100	98	86
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	143	96	96	96
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614	300169	45	100	95	78
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631	103000	119	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
AK2	Reticular dysgenesis, 267500	103020	69	100	100	92
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555	604741	54	100	100	91
ALAD	{Lead poisoning, susceptibility to}, 612740 Porphyria, acute hepatic, 612740	125270	96	100	100	100
ALAS2	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, 300752	301300	51	100	98	87
ALDH18A1	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586	138250	68	100	100	95
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	82	100	100	92
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	58	100	98	92
ALDH4A1	Hyperprolinemia, type II, 239510	606811	115	100	100	100
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	61	100	97	91
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	603178	94	100	100	95
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	60	100	98	81
ALDOA	Glycogen storage disease XII, 611881	103850	134	100	100	100
ALDOB	Fructose intolerance, hereditary, 229600	612724	89	100	100	100
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	59	91	79	72
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	61	100	100	97
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	152	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	41	100	93	68
ALG14	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227	612866	85	100	100	88
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	70	100	100	97
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	78	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	63	100	100	90
ALG8	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874	608103	54	100	99	88
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	54	100	99	89
ALOX12B	Ichthyosis, congenital, autosomal recessive 2, 242100	603741	106	100	100	99
ALPL	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300	171760	113	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
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	80	100	100	96
AMN	Megaloblastic anemia-1, Norwegian type, 261100	605799	90	100	96	85
AMPD1	Myopathy due to myoadenylate deaminase deficiency, 615511	102770	58	100	98	89
AMT	Glycine encephalopathy, 605899	238310	108	100	100	100
APOA1	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined Corneal clouding, autosomal recessive Hypoalphalipoproteinemia, 604091	107680	134	100	100	100
APOB	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558	107730	143	100	100	99
APOC2	Hyperlipoproteinemia, type Ib, 207750	608083	79	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	103	100	100	97
APRT	Adenine phosphoribosyltransferase deficiency, 614723	102600	86	100	100	100
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	67	100	98	87
ARG1	Argininemia, 207800	608313	63	100	100	92
ARSA	Metachromatic leukodystrophy, 250100	607574	136	100	100	100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	611542	68	100	100	95
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	57	100	98	85
ASL	Argininosuccinic aciduria, 207900	608310	110	100	100	98
ASNS	Asparagine synthetase deficiency, 615574	108370	64	100	99	90
ASPA	Canavan disease, 271900	608034	49	100	98	86
ASS1	Citrullinemia, 215700	603470	111	100	97	88
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	54	100	99	93
ATP5F1E	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	606153	81	100	100	100
ATP6AP1	Immunodeficiency 47, 300972	300197	70	100	99	96
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	70	100	99	94
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489	300011	40	100	95	71
ATP7B	Wilson disease, 277900	606882	88	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
ATP8B1	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600	602397	60	100	98	89
ATPAF2	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	68	100	100	98
AUH	3-methylglutaconic aciduria, type I, 250950	600529	84	100	100	95
B3GALNT1	[Blood group, P1PK system, P(k) phenotype], 111400 [Blood group, globoside system], 615021	603094	78	100	100	100
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	52	100	99	89
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	66	79	75	72
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	60	100	95	79
B4GALNT1	Spastic paraplegia 26, autosomal recessive, 609195	601873	108	100	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	98	100	100	98
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	604327	129	100	100	98
BAAT	Hypercholanemia, familial, 607748	602938	62	100	97	83
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	146	100	100	99
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	56	100	98	89
BCO1	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300	605748	63	100	100	94
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	165	100	100	100
BLVRA	Hyperbiliverdinemia, 614156	109750	70	100	100	96
BMP2	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877	112261	112	100	100	98
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299	613183	67	100	99	82
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800	613896	59	100	100	96
BTD	Biotinidase deficiency, 253260	609019	82	100	100	98
C1GALT1C1	Tn polyagglutination syndrome, somatic, 300622	300611	40	100	100	85
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	164	100	100	100
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	109	100	100	100
CANT1	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719	613165	124	100	100	100
CAT	Acatalasemia, 614097	115500	61	100	98	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	122	100	100	100
CCDC115	Congenital disorder of glycosylation, type IIo, 616828	613734	63	85	82	79
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646	606475	90	100	100	99
CEL	Maturity-onset diabetes of the young, type VIII, 609812	114840	145	97	93	91
CERKL	Retinitis pigmentosa 26, 608380	608381	63	100	99	89
CERS3	Ichthyosis, congenital, autosomal recessive 9, 615023	615276	57	100	99	91
CETP	[High density lipoprotein cholesterol level QTL 10], 143470 Hyperalphalipoproteinemia, 143470	118470	86	100	100	100
CHKB	Muscular dystrophy, congenital, megaconial type, 602541	612395	104	100	100	100
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1, 601776	608429	123	100	98	97
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	603799	120	100	100	100
CHST6	Macular corneal dystrophy, 217800	605294	240	100	100	100
CHSY1	Temtamy preaxial brachydactyly syndrome, 605282	608183	71	99	96	92
CLDN16	Hypomagnesemia 3, renal, 248250	603959	64	100	100	95
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	143	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	101	100	100	99
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	62	100	100	99
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	117	100	100	99
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	123	100	100	100
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	110	100	100	99
CLPS	No OMIM phenotype	120105	98	100	100	100
CLPX	?Protoporphyria, erythropoietic, 2, 618015	615611	47	100	95	74
CNDP1	No OMIM phenotype	609064	64	100	99	92
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	148	100	100	97
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	89	100	99	95
COG2	?Congenital disorder of glycosylation, type IIq, 617395	606974	56	100	99	89
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	75	100	100	96
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	58	100	98	87
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	58	100	98	82
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	77	100	100	96
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	120	100	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	99	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	125	100	100	99
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	131	100	100	96
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	56	100	97	87
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	87	100	100	100
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	54	100	99	89
CPOX	Coproporphyria, 121300 Harderoporphyria, 121300	612732	69	100	100	93
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	52	100	99	91
CPT1A	CPT deficiency, hepatic, type IA, 255120	600528	100	100	100	97
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	93	100	100	99
CTH	Cystathioninuria, 219500 Homocysteine, total plasma, elevated	607657	66	100	100	94
CTNS	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750	606272	112	100	100	100
CTSA	Galactosialidosis, 256540	613111	118	100	100	100
CTSC	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650	602365	64	100	100	94
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	138	100	100	100
CTSK	Pycnodysostosis, 265800	601105	51	100	100	95
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	71	100	99	90
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	120	100	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	126	100	100	100
CYP27B1	Vitamin D-dependent rickets, type I, 264700	609506	109	100	100	100
CYP7A1	No OMIM phenotype	118455	59	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800	603711	60	100	100	96
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	148	100	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	53	100	100	92
DBH	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]	609312	126	100	100	100
DBT	Maple syrup urine disease, type II, 248600	248610	99	100	100	98
DCXR	[Pentosuria], 260800	608347	156	100	100	100
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	75	100	97	89
DDHD1	Spastic paraplegia 28, autosomal recessive, 609340	614603	96	100	98	88
DDOST	?Congenital disorder of glycosylation, type Ir, 614507	602202	85	100	100	100
DGAT1	?Diarrhea 7, 615863	604900	108	97	91	88
DGKE	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008	601440	92	100	100	93
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070	601465	67	100	99	83
DHCR24	Desmosterolosis, 602398	606418	113	100	100	100
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	98	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	61	100	100	99
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	74	100	100	97
DHODH	Miller syndrome, 263750	126064	83	100	100	92
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	76	100	100	96
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	70	100	96	83
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	66	100	100	94
DLST	No OMIM phenotype	126063	72	100	100	98
DMGDH	Dimethylglycine dehydrogenase deficiency, 605850	605849	64	100	100	96
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	60	100	94	85
DNAJC19	3-methylglutaconic aciduria, type V, 610198	608977	65	100	100	89
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	218	100	100	100
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708	603850	61	100	98	89
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116	126375	87	100	99	95
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	123	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
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	66	100	100	97
DPEP1	No OMIM phenotype	179780	115	100	100	100
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	70	93	89	83
DPM2	Congenital disorder of glycosylation, type Iu, 615042	603564	67	100	100	96
DPM3	Congenital disorder of glycosylation, type Io, 612937	605951	227	100	100	100
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	54	100	99	90
DPYS	Dihydropyrimidinuria, 222748	613326	63	100	100	96
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960	300205	76	100	100	100
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277	602292	74	100	100	98
EGF	Hypomagnesemia 4, renal, 611718	131530	63	100	99	92
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	61	100	100	93
ENO3	?Glycogen storage disease XIII, 612932	131370	138	100	100	100
EOGT	Adams-Oliver syndrome 4, 615297	614789	50	100	99	86
EPG5	Vici syndrome, 242840	615068	61	100	99	93
EPHX1	?Hypercholanemia, familial, 607748	132810	120	100	100	99
EPHX2	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890	132811	80	100	100	92
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	76	89	87	84
ETFA	Glutaric acidemia IIA, 231680	608053	53	100	100	92
ETFB	Glutaric acidemia IIB, 231680	130410	102	100	100	100
ETFDH	Glutaric acidemia IIC, 231680	231675	73	100	100	97
ETHE1	Ethylmalonic encephalopathy, 602473	608451	108	100	100	93
EXT1	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700	608177	63	100	98	92
EXT2	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	89	100	99	94
FA2H	Spastic paraplegia 35, autosomal recessive, 612319	611026	73	100	100	95
FAH	Tyrosinemia, type I, 276700	613871	91	100	100	98
FASTKD2	?Mitochondrial complex IV deficiency, 220110	612322	65	100	99	92
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700	611570	121	100	100	100
FECH	Protoporphyria, erythropoietic, 1, 177000	612386	57	100	100	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579	176943	63	100	99	90
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	70	99	94	86
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	130	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	72	100	100	98
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100	610595	124	100	100	100
FMO3	Trimethylaminuria, 602079	136132	59	100	99	92
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	96	100	100	100
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	613622	85	100	100	100
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	93	99	96	93
FUCA1	Fucosidosis, 230000	612280	79	100	100	94
FXYD2	Hypomagnesemia 2, renal, 154020	601814	89	100	100	100
G6PC	Glycogen storage disease Ia, 232200	613742	96	100	100	100
G6PC3	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541	611045	96	100	100	100
G6PD	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162	305900	90	100	100	99
GAA	Glycogen storage disease II, 232300	606800	140	100	100	100
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513	605363	78	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GALC	Krabbe disease, 245200	606890	47	100	97	82
GALE	Galactose epimerase deficiency, 230350	606953	109	100	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	133	100	100	98
GALNS	Mucopolysaccharidosis IVA, 253000	612222	76	100	98	93
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900	601756	60	100	100	95
GALT	Galactosemia, 230400	606999	146	100	100	100
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	80	100	99	95
GANAB	Polycystic kidney disease 3, 600666	104160	71	100	99	94
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	56	100	100	93
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	173	100	100	100
GBA2	Spastic paraplegia 46, autosomal recessive, 614409	609471	129	100	100	100
GBE1	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	607839	55	100	97	83
GCDH	Glutaricaciduria, type I, 231670	608801	104	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	49	100	100	91
GCK	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851	138079	117	100	100	100
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446	606857	76	100	100	93
GCSH	?Glycine encephalopathy, 605899	238330	88	100	91	63
GFM1	Combined oxidative phosphorylation deficiency 1, 609060	606639	61	100	99	92
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, 610542	138292	49	100	98	84
GGT1	?Glutathioninuria, 231950	612346	59	77	64	59
GIF	Intrinsic factor deficiency, 261000	609342	72	100	100	96
GK	Glycerol kinase deficiency, 307030	300474	40	99	86	60
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	88	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	96	100	100	96
GLDC	Glycine encephalopathy, 605899	238300	61	100	97	87
GLRA1	Hyperekplexia 1, 149400	138491	77	100	99	93
GLRX5	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859	609588	66	100	91	81
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	89	99	96	88
GLUL	Glutamine deficiency, congenital, 610015	138290	82	100	100	100
GLYCTK	D-glyceric aciduria, 220120	610516	135	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	85	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	99	100	100	99
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352	615320	168	100	100	100
GNE	Nonaka myopathy, 605820 Sialuria, 269921	603824	80	100	100	97
GNMT	Glycine N-methyltransferase deficiency, 606664	606628	109	100	100	100
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	66	100	99	91
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	54	100	98	89
GNPTG	Mucolipidosis III gamma, 252605	607838	149	100	100	95
GNS	Mucopolysaccharidosis type IIID, 252940	607664	60	100	100	96
GPD1	Hypertriglyceridemia, transient infantile, 614480	138420	71	100	100	99
GPHN	Molybdenum cofactor deficiency C, 615501	603930	59	100	100	93
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470	172400	117	100	100	100
GPX1	Hemolytic anemia due to glutathione peroxidase deficiency, 614164	138320	79	100	100	100
GRHPR	Hyperoxaluria, primary, type II, 260000	604296	84	100	100	100
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	82	100	100	98
GSTZ1	[Maleylacetoacetate isomerase deficiency], 617596	603758	94	100	100	100
GUSB	Mucopolysaccharidosis VII, 253220	611499	100	100	100	97
GYG1	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199	603942	51	100	100	85
GYS1	Glycogen storage disease 0, muscle, 611556	138570	115	100	100	100
GYS2	Glycogen storage disease 0, liver, 240600	138571	54	100	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	67	100	100	98
HADHA	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015	600890	90	100	100	93
HADHB	Trifunctional protein deficiency, 609015	143450	58	100	100	91
HAL	[Histidinemia], 235800	609457	79	100	100	95
HAMP	Hemochromatosis, type 2B, 613313	606464	154	100	100	100
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	71	100	98	89
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	79	100	100	96
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	106	100	100	95
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	81	100	100	97
HGD	Alkaptonuria, 203500	607474	54	100	99	91
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	60	94	94	88
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	610690	45	100	91	67
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	93	100	99	98
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	85	100	100	97
HMBS	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000	609806	82	100	100	100
HMGCL	HMG-CoA lyase deficiency, 246450	613898	78	100	100	100
HMGCS2	HMG-CoA synthase-2 deficiency, 605911	600234	69	100	100	97
HMOX1	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	141250	102	100	100	100
HOGA1	Hyperoxaluria, primary, type III, 613616	613597	125	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	94	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	34	100	93	70
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	97	100	100	97
HS6ST1	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880	604846	145	100	100	99
HSD11B1	Cortisone reductase deficiency 2, 614662	600713	54	100	100	96
HSD11B2	Apparent mineralocorticoid excess, 218030	614232	136	92	87	83
HSD17B10	HSD10 mitochondrial disease, 300438	300256	62	100	100	100
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	54	100	96	87
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810	613890	122	100	100	100
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765	607764	110	100	100	100
HYAL1	?Mucopolysaccharidosis type IX, 601492	607071	99	100	100	100
IBA57	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451	615316	126	100	100	98
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	99	100	100	99
IDH3B	Retinitis pigmentosa 46, 612572	604526	97	100	100	100
IDS	Mucopolysaccharidosis II, 309900	300823	68	100	98	84
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	127	100	99	94
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type, 614078	614010	125	100	99	94
IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	99	100	99	94
INPPL1	Opsismodysplasia, 258480	600829	101	100	100	99
ISCU	Myopathy with lactic acidosis, hereditary, 255125	611911	72	100	100	97
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	77	100	99	92
ITPA	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850	147520	115	100	100	100
IVD	Isovaleric acidemia, 243500	607036	105	100	100	98
KHK	[Fructosuria], 229800	614058	116	100	100	99
KYNU	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661	605197	64	100	100	91
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	61	100	100	93
LAMP2	Danon disease, 300257	309060	40	100	91	68
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840	603590	86	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
LBR	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471	600024	57	100	99	92
LCAT	Fish-eye disease, 136120 Norum disease, 245900	606967	124	100	100	97
LCT	Lactase deficiency, congenital, 223000	603202	103	100	100	100
LDHA	Glycogen storage disease XI, 612933	150000	51	100	97	79
LDLR	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890	606945	207	100	100	100
LFNG	?Spondylocostal dysostosis 3, autosomal recessive, 609813	602576	112	86	83	82
LIPA	Cholesteryl ester storage disease, 278000 Wolman disease, 278000	613497	58	100	100	93
LIPC	{Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797	151670	73	100	100	96
LIPI	{Hypertriglyceridemia, susceptibility to}, 145750	609252	52	100	98	84
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380	612625	57	100	96	76
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive, 268200	605518	72	100	99	93
LPIN2	Majeed syndrome, 609628	605519	65	100	99	93
LPL	Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11] Lipoprotein lipase deficiency, 238600	609708	102	100	100	98
LRP2	Donnai-Barrow syndrome, 222448	600073	58	100	99	91
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	51	100	98	84
LTC4S	Leukotriene C4 synthase deficiency, 614037	246530	70	100	87	76
LYST	Chediak-Higashi syndrome, 214500	606897	62	100	98	91
MAN1B1	Mental retardation, autosomal recessive 15, 614202	604346	120	100	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	110	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	76	100	98	92
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	42	100	97	83
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850	610550	100	100	100	100
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	609010	60	100	99	89
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	56	100	99	90
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120	608419	69	100	100	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MCOLN1	Mucolipidosis IV, 252650	605248	121	100	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	56	100	99	89
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	94	100	100	100
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	69	100	98	90
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	67	100	100	97
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110	607568	78	100	100	96
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	115	100	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	55	100	99	81
MOCOS	Xanthinuria, type II, 603592	613274	76	100	99	92
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	108	100	100	99
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	60	100	100	94
MOCS3	No OMIM phenotype	609277	175	100	100	100
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	117	100	100	100
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	78	100	100	98
MPI	Congenital disorder of glycosylation, type Ib, 602579	154550	161	100	100	98
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	137960	92	100	100	98
MRPS16	Combined oxidative phosphorylation deficiency 2, 610498	609204	101	100	100	100
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	70	100	100	91
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834	607545	86	100	100	96
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	172460	67	100	99	91
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	95	100	100	99
MTM1	Myotubular myopathy, X-linked, 310400	300415	37	100	88	65
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	69	100	99	93
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	81	100	99	96
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	55	100	99	92
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	62	100	99	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	96	100	100	98
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	114	100	100	99
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	114	100	97	93
NAGS	N-acetylglutamate synthase deficiency, 237310	608300	102	100	100	100
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	57	100	99	93
NAT8L	?N-acetylaspartate deficiency, 614063	610647	68	95	90	85
NDUFA1	Mitochondrial complex I deficiency, 252010	300078	99	100	100	100
NDUFA11	Mitochondrial complex I deficiency, 252010	612638	115	100	100	100
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	98	100	100	100
NDUFAF1	Mitochondrial complex I deficiency, 252010	606934	53	100	100	90
NDUFAF2	Mitochondrial complex I deficiency, 252010	609653	103	100	88	71
NDUFAF3	Mitochondrial complex I deficiency, 252010	612911	125	100	100	100
NDUFAF4	Mitochondrial complex I deficiency, 252010	611776	83	100	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010	612360	63	100	99	89
NDUFS1	Mitochondrial complex I deficiency, 252010	157655	61	100	99	87
NDUFS2	Mitochondrial complex I deficiency, 252010	602985	71	100	100	96
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	603846	102	100	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010	602694	69	100	100	97
NDUFS6	Mitochondrial complex I deficiency, 252010	603848	69	100	100	97
NDUFS7	Leigh syndrome, 256000	601825	114	100	100	99
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	131	100	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010	161015	122	100	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010	600532	45	100	93	75
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	143	100	100	100
NFU1	Multiple mitochondrial dysfunctions syndrome 1, 605711	608100	58	100	100	90
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	130	100	100	100
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	74	100	100	97
NPC2	Niemann-pick disease, type C2, 607625	601015	87	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	62	100	98	88
NT5C	No OMIM phenotype	191720	90	100	88	72
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency, 266120	606224	55	100	98	81
NT5E	Calcification of joints and arteries, 211800	129190	80	100	100	96
NUBPL	Mitochondrial complex I deficiency, 252010	613621	51	100	100	93
NUS1	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831	610463	53	100	98	87
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	54	100	94	80
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	35	100	91	67
OGDH	Alpha-ketoglutarate dehydrogenase deficiency, 203740	613022	119	100	100	98
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	53	100	96	83
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	124	100	100	100
OPLAH	5-oxoprolinase deficiency, 260005	614243	120	100	100	99
OTC	Ornithine transcarbamylase deficiency, 311250	300461	46	100	94	80
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	601424	68	100	100	92
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	57	100	98	91
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	72	100	100	96
PC	Pyruvate carboxylase deficiency, 266150	608786	135	100	100	99
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070	126090	75	100	100	94
PCCA	Propionicacidemia, 606054	232000	58	100	99	90
PCCB	Propionicacidemia, 606054	232050	62	100	97	92
PCK1	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680	614168	105	100	100	100
PCK2	PEPCK deficiency, mitochondrial, 261650	614095	115	100	100	100
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	41	99	93	77
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	59	100	100	90
PDHX	Lacticacidemia due to PDX1 deficiency, 245349	608769	65	100	100	95
PDK1	No OMIM phenotype	602524	72	100	99	91
PDK2	No OMIM phenotype	602525	103	100	100	100
PDK3	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905	300906	44	100	91	67

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PDK4	No OMIM phenotype	602527	48	100	99	86
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	76	100	100	100
PDP2	No OMIM phenotype	615499	110	100	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	51	99	94	83
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	64	100	95	81
PEPD	Prolidase deficiency, 170100	613230	92	100	100	97
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	52	100	98	90
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	96	100	100	96
PEX11B	?Peroxisome biogenesis disorder 14B, 614920	603867	136	100	100	100
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	57	100	99	87
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	58	100	100	95
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887	601791	123	100	100	100
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	124	100	96	93
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	56	100	100	94
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	57	100	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	123	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	50	100	99	90
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	104	100	100	99
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	97	100	100	96
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	49	100	100	95
PFKM	Glycogen storage disease VII, 232800	610681	83	100	100	98
PGAM2	Glycogen storage disease X, 261670	612931	161	100	100	100
PGAP1	Mental retardation, autosomal recessive 42, 615802	611655	56	100	97	83
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	141	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	107	100	100	100
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	37	100	99	81
PGM1	Congenital disorder of glycosylation, type It, 614921	171900	61	100	100	96
PGM3	Immunodeficiency 23, 615816	172100	75	100	100	94
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	135	100	100	99
PHKA1	Muscle glycogenosis, 300559	311870	40	100	93	69
PHKA2	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000	300798	47	100	97	84
PHKB	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750	172490	65	100	100	95
PHKG1	No OMIM phenotype	172470	111	100	100	95
PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027	172471	119	100	100	100
PHYH	Refsum disease, 266500	602026	109	100	98	87
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	56	100	100	93
PIGC	Glycosylphosphatidylinositol biosynthesis defect 16, 617816	601730	122	100	100	100
PIGG	Mental retardation, autosomal recessive 53, 616917	616918	92	100	100	98
PIGL	CHIME syndrome, 280000	605947	87	100	100	97
PIGM	Glycosylphosphatidylinositol deficiency, 610293	610273	77	100	100	100
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	53	100	97	80
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	112	100	100	100
PIGQ	No OMIM phenotype	605754	163	100	100	100
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	124	100	100	100
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	87	100	100	99
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11, 616025	610275	75	100	100	100
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	42	100	100	92
PIK3R1	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	72	100	100	94
PIK3R5	Ataxia-oculomotor apraxia 3, 615217	611317	101	100	100	99
PKLR	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200	609712	145	100	100	100
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953	603604	115	100	100	100
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	51	100	99	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PLIN1	Lipodystrophy, familial partial, type 4, 613877	170290	95	100	100	99
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400	153454	98	100	100	99
PLOD2	Bruck syndrome 2, 609220	601865	48	100	96	81
PLOD3	Lysyl hydroxylase 3 deficiency, 612394	603066	97	100	100	99
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	66	100	100	93
PMPCA	Spinocerebellar ataxia, autosomal recessive 2, 213200	613036	133	100	100	100
PNLIP	?Pancreatic lipase deficiency, 614338	246600	52	100	98	89
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	61	100	99	94
PNPLA2	Neutral lipid storage disease with myopathy, 610717	609059	106	100	100	100
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020	603197	113	100	100	98
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	69	100	100	94
POFUT1	Dowling-Degos disease 2, 615327	607491	103	100	100	100
POGLUT1	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, type 2Z, 617232	615618	45	100	99	88
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450	174763	103	100	100	98
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131	604983	121	100	99	93
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	79	100	100	94
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	66	100	98	88
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123	606822	80	100	100	98
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830	614828	145	100	100	100
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094	615247	80	100	100	100
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	96	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	78	100	100	95
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	124015	152	100	100	100
PPM1K	?Maple syrup urine disease, mild variant, 615135	611065	68	100	100	93
PPOX	Porphyria variegata, 176200	600923	103	100	100	99
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	75	100	100	97
PRKAG2	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200	602743	111	100	100	97
PRKCSH	Polycystic liver disease 1, 174050	177060	125	100	100	99
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	96	100	96	88
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500, X-linked Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	44	100	96	81
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	122	100	100	97
PSAT1	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	610936	57	100	100	91
PSPH	Phosphoserine phosphatase deficiency, 614023	172480	48	100	94	78
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	75	100	100	90
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	86	100	100	95
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438	179035	96	100	100	96
PYGL	Glycogen storage disease VI, 232700	613741	75	100	99	90
PYGM	McArdle disease, 232600	608455	109	100	100	100
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	80	100	100	95
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	57	100	97	85
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895	610924	132	100	100	100
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	121	100	100	100
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	55	100	98	88

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	98	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	54	100	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	279	100	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	100	100	100	94
RPIA	?Ribose 5-phosphate isomerase deficiency, 608611	180430	70	100	100	96
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, autosomal dominant 5, 613077	604712	79	100	100	98
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	91	100	98	87
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	58	100	97	80
SAR1B	Chylomicron retention disease, 246700	607690	67	100	100	93
SARDH	[Sarcosinemia], 268900	604455	100	100	100	99
SC5D	Lathosterolosis, 607330	602286	70	100	100	98
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	602257	78	100	99	93
SCO1	Mitochondrial complex IV deficiency, 220110	603644	97	100	100	97
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	126	100	100	100
SCP2	?Leukoencephalopathy with dystonia and motor neuropathy, 613724	184755	47	100	99	87
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	128	100	97	90
SDHAF1	Mitochondrial complex II deficiency, 252011	612848	83	100	100	100
SDHB	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300	185470	138	100	100	100
SDHC	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373	602413	162	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
SDHD	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300	602690	155	100	100	100
SEC23B	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100	610512	62	100	99	91
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	65	100	99	91
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	56	100	95	79
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	102	100	95	89
SI	Sucrase-isomaltase deficiency, congenital, 222900	609845	50	100	97	83
SLC12A3	Gitelman syndrome, 263800	600968	105	100	100	99
SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095	600682	74	100	100	97
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	73	100	100	94
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270	603941	58	100	100	96
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	61	100	100	96
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	84	100	100	95
SLC1A3	Episodic ataxia, type 6, 612656	600111	90	100	100	100
SLC22A5	Carnitine deficiency, systemic primary, 212140	603377	97	100	100	99
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182	190315	100	100	100	100
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	79	100	100	96
SLC25A13	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814	603859	87	100	98	87
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	121	100	100	96
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	99	100	100	99
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138	613698	54	100	100	92
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	111	100	100	100
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773	600370	90	100	100	98
SLC25A32	?Exercise intolerance, riboflavin-responsive, 616839	610815	51	100	93	69
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950	610819	103	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
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, autosomal dominant 2, 609283	103220	96	100	100	100
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	117	100	100	100
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi-Bickel syndrome, 227810	138160	54	100	99	92
SLC30A10	Hypermanganesemia with dystonia 1, 613280	611146	127	100	100	99
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539	603690	60	100	96	81
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	60	100	100	93
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	62	100	96	84
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	133	100	100	100
SLC35D1	Schneckenbecken dysplasia, 269250	610804	51	100	97	77
SLC36A2	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608331	92	100	100	99
SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	87	100	99	93
SLC39A14	Hypermanganesemia with dystonia 2, 617013 ?Hyperostosis cranalis interna, 144755	608736	85	100	99	95
SLC39A4	Acrodermatitis enteropathica, 201100	607059	122	100	100	100
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721	608732	67	100	99	90
SLC3A1	Cystinuria, 220100	104614	79	100	99	94
SLC40A1	Hemochromatosis, type 4, 606069	604653	54	100	99	93
SLC46A1	Folate malabsorption, hereditary, 229050	611672	113	100	100	100
SLC52A1	Riboflavin deficiency, 615026	607883	180	100	100	100
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	166	100	100	100
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500	613350	103	100	100	100
SLC5A1	Glucose/galactose malabsorption, 606824	182380	94	100	99	93
SLC5A2	Renal glucosuria, 233100	182381	108	100	100	100
SLC6A19	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608893	109	100	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	91	100	99	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SLC7A7	Lysinuric protein intolerance, 222700	603593	67	100	99	94
SLC7A9	Cystinuria, 220100	604144	92	100	100	98
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic, 237450	604843	53	100	98	83
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic, 237450	605495	58	100	96	88
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	130	100	100	98
SOD1	Amyotrophic lateral sclerosis 1, 105400	147450	67	100	100	99
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	100	100	100	100
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA, 162400	605712	57	100	98	91
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640	605713	71	100	99	90
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	107	100	99	95
SSR3	No OMIM phenotype	606213	59	92	76	55
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	73	100	100	98
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090	606494	71	100	99	94
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	47	99	90	77
STAR	Lipoid adrenal hyperplasia, 201710	600617	106	100	100	100
STS	Ichthyosis, X-linked, 308100	300747	77	97	97	91
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	52	100	99	95
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	79	100	99	93
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	51	100	96	81
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	82	100	100	99
SUGCT	Glutaric aciduria III, 231690	609187	64	100	98	90
SUMF1	Multiple sulfatase deficiency, 272200	607939	83	100	100	96
SUOX	Sulfite oxidase deficiency, 272300	606887	124	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	78	95	89	87
TACO1	Mitochondrial complex IV deficiency, 220110	612958	73	100	100	100
TALDO1	Transaldolase deficiency, 606003	602063	128	100	100	100
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	616830	87	100	100	99
TAT	Tyrosinemia, type II, 276600	613018	54	100	100	94
TAZ	Barth syndrome, 302060	300394	89	100	97	90
TBXAS1	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158	274180	68	100	98	90
TCIRG1	Osteopetrosis, autosomal recessive 1, 259700	604592	116	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
TCN1	No OMIM phenotype	189905	53	100	98	90
TCN2	Transcobalamin II deficiency, 275350	613441	107	100	100	100
TDO2	[?Hypertryptophanemia], 600627	191070	42	100	97	81
TECR	Mental retardation, autosomal recessive 14, 614020	610057	135	100	100	99
TFR2	Hemochromatosis, type 3, 604250	604720	92	100	100	97
TH	Segawa syndrome, recessive, 605407	191290	92	100	99	94
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	131	100	100	100
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069	188250	58	100	100	95
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	109	100	100	98
TMEM199	Congenital disorder of glycosylation, type IIp, 616829	616815	78	100	100	99
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	81	100	100	93
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	190450	112	100	99	96
TPMT	{Thiopurines, poor metabolism of, 1}, 610460	187680	73	100	100	81
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	87	100	100	99
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	51	100	96	84
TREH	Trehalase deficiency, 612119	275360	104	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	217	100	100	100
TRIM37	Mulibrey nanism, 253250	605073	52	100	98	87
TRMU	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070	610230	92	100	100	97
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	62	100	99	92
TSFM	Combined oxidative phosphorylation deficiency 3, 610505	604723	70	100	100	100
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	46	100	93	72
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	54	100	97	87
TUFM	Combined oxidative phosphorylation deficiency 4, 610678	602389	151	100	100	100
TUSC3	Mental retardation, autosomal recessive 7, 611093	601385	64	100	100	91
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286	606075	130	100	100	100
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	131222	92	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
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	77	100	100	96
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	65	100	100	96
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	111	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	85	100	100	93
UMPS	Orotic aciduria, 258900	613891	63	100	100	95
UPB1	Beta-ureidopropionase deficiency, 613161	606673	91	100	100	99
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158	191330	69	100	100	100
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	120	100	100	100
UROC1	?Urocanase deficiency, 276880	613012	107	100	100	99
UROD	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100	613521	77	100	100	94
UROS	Porphyria, congenital erythropoietic, 263700	606938	57	100	100	93
USF1	{Hyperlipidemia, familial combined, susceptibility to}, 602491	191523	110	100	100	100
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700	608547	94	100	100	100
VPS13B	Cohen syndrome, 216550	607817	63	100	98	91
XDH	Xanthinuria, type I, 278300	607633	71	100	99	93
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	97	100	97	92
XYLT2	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822	608125	128	100	100	100
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	610957	117	100	100	98

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors

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

- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 95 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x