Whole Exome Sequencing
Gene package Metabolic disorders, version 1.1, 22-11-2017

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

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
AASS	Hyperlysinemia, 238700Saccharopinuria, 268700	605113	45	100	95	80
ABAT	GABA-transaminase deficiency, 613163	137150	65	100	98	87
ABCA1	{Coronary artery disease in familial hypercholesterolemia, protection against}, 143890 HDL deficiency, type 2, 604091 Tangier disease, 205400	600046	62	100	97	88
ABCB11	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847	603201	46	100	97	83
ABCB4	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803	171060	48	100	98	80
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	53	100	95	83
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	56	83	72	70
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	603214	71	100	100	94
ABCG5	Sitosterolemia, 210250	605459	53	100	99	93
ABCG8	{Gallbladder disease 4}, 611465 Sitosterolemia, 210250	605460	93	100	100	98
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283	604773	97	100	100	100
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126	611103	64	100	100	98

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ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	607008	55	100	99	90
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	606885	84	100	100	98
ACADSB	2-methylbutyrylglycinuria, 610006	600301	48	100	95	73
ACADVL	VLCAD deficiency, 201475	609575	81	100	98	93
ACAT1	Alpha-methylacetoacetic aciduria, 203750	607809	51	100	99	84
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	71	100	100	94
ACSF3	Combined malonic and methylmalonic aciduria, 614265	614245	86	100	100	96
ACY1	Aminoacylase 1 deficiency, 609924	104620	72	100	100	100
ADA	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700	608958	57	100	98	85
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300	102750	38	100	90	62
ADSL	Adenylosuccinase deficiency, 103050	608222	61	100	99	88
AGA	Aspartylglucosaminuria, 208400	613228	53	100	98	85
AGK	Cataract 38, 614691 Sengers syndrome, 212350	610345	44	100	98	83
AGL	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400	610860	53	100	99	89
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121	603051	48	100	96	74
AGXT	Hyperoxaluria, primary, type 1, 259900	604285	79	100	100	100
AGXT2	[Beta-aminoisobutyric acid, urinary excretion of], 210100	612471	47	100	96	79
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	180960	93	96	96	95
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness 5, 300614	300169	41	100	89	67
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555	604741	48	100	92	86
ALAD	{Lead poisoning, susceptibility to}, 612740 Porphyria, acute hepatic, 612740	125270	71	100	100	97
ALAS2	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, 300752	301300	45	100	97	84
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	54	100	98	88
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	52	100	98	88
ALDH4A1	Hyperprolinemia, type II, 239510	606811	64	100	99	97
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	42	99	94	71
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	603178	71	100	99	93
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	50	100	93	71

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ALDOA	Glycogen storage disease XII, 611881	103850	95	100	100	100
ALDOB	Fructose intolerance, 229600	612724	66	100	100	100
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	46	90	76	70
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	59	100	100	99
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	108	100	100	100
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	42	100	96	73
ALG14	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227	612866	69	100	100	91
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	607905	51	100	100	97
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	63	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	58	100	99	83
ALG8	Congenital disorder of glycosylation, type Ih, 608104	608103	43	100	96	75
ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	43	100	96	77
ALPL	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300	171760	64	100	100	96
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	53	100	100	89
AMN	Megaloblastic anemia-1, Norwegian type, 261100	605799	57	100	99	82
AMPD1	Myopathy due to myoadenylate deaminase deficiency, 615511	102770	48	100	97	85
AMT	Glycine encephalopathy, 605899	238310	73	100	100	98
APOA1	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined Corneal clouding Hypoalphalipoproteinemia, 604091	107680	86	100	100	100
APOB	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558	107730	115	100	100	99
APOC2	Hyperlipoproteinemia, type Ib, 207750	608083	62	100	100	100
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	76	100	100	95
APRT	Adenine phosphoribosyltransferase deficiency, 614723	102600	52	100	100	100

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APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	54	100	90	77
ARG1	Argininemia, 207800	608313	56	100	100	88
ARSA	Metachromatic leukodystrophy, 250100	607574	105	100	100	100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	611542	49	100	99	90
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	49	100	97	80
ASL	Argininosuccinic aciduria, 207900	608310	70	100	100	99
ASNS	Asparagine synthetase deficiency, 615574	108370	59	100	100	92
ASPA	Canavan disease, 271900	608034	41	100	98	81
ASS1	Citrullinemia, 215700	603470	70	100	100	98
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688	601731	45	100	99	84
ATP5E	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	606153	56	100	100	100
ATP6AP1	Immunodeficiency 47, 300972	300197	58	100	96	86
ATP6V0A2	Cutis laxa, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	56	100	98	87
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	44	100	98	84
ATP7B	Wilson disease, 277900	606882	70	100	100	97
ATP8B1	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600	602397	50	100	98	85
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	608918	51	100	100	90
AUH	3-methylglutaconic aciduria, type I, 250950	600529	52	100	100	92
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	42	100	97	76
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	48	79	76	72
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600	606374	73	100	100	100
B3GLCT	Peters-plus syndrome, 261540	610308	47	100	87	64
B4GALNT1	Spastic paraplegia 26, 609195	601873	71	100	100	100
B4GALT1	Congenital disorder of glycosylation, type IId, 607091	137060	69	100	100	94
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070	604327	76	100	100	97
BAAT	Hypercholanemia, familial, 607748	602938	57	100	98	83
BCAT1	?Hyperleucinemia-isoleucinemia or hypervalinemia	113520	40	100	92	68
BCAT2	?Hypervalinemia or hyperleucine-isoleucinemia	113530	62	100	100	96
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	86	100	100	100
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	49	100	99	86

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BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	134	100	100	100
BTD	Biotinidase deficiency, 253260	609019	70	100	100	98
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	114761	110	100	100	100
CAD	Epileptic encephalopathy, early infantile, 50, 616457	114010	76	100	100	100
CAT	Acatalasemia, 614097	115500	46	100	97	81
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	70	100	100	96
CCDC115	Congenital disorder of glycosylation, type IIo, 616828	613734	39	84	77	72
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646	606475	65	100	100	96
CETP	[High density lipoprotein cholesterol level QTL 10], 143470 Hyperalphalipoproteinemia, 143470	118470	61	100	100	99
CHSY1	Temtamy preaxial brachydactyly syndrome, 605282	608183	61	98	95	93
CLDN16	Hypomagnesemia 3, renal, 248250	603959	51	100	99	89
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	75	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	76	100	100	94
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	57	100	100	98
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	78	100	98	88
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	81	100	100	100
CLPS	No OMIM phenotype	120105	77	100	100	100
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	106	100	100	95
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	66	100	99	94
COG2	?Congenital disorder of glycosylation, type IIq, 617395	606974	47	100	98	81
COG4	Congenital disorder of glycosylation, type IIj, 613489	606976	53	100	100	89
COG5	Congenital disorder of glycosylation, type IIi, 613612	606821	43	100	96	77
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	48	100	96	74
COG7	Congenital disorder of glycosylation, type IIe, 608779	606978	56	100	98	85
COG8	Congenital disorder of glycosylation, type IIh, 611182	606979	80	100	100	99
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	49	100	97	82
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	86	100	100	100

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COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	43	100	96	76
COX6B1	Mitochondrial complex IV deficiency, 220110	124089	60	100	100	100
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	47	100	99	87
CPOX	Coproporphyria, 121300 Harderoporphyria, 121300	612732	46	100	99	85
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	42	100	96	78
CPT2	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212	600650	72	100	100	98
CTH	Cystathioninuria, 219500 Homocysteine, total plasma, elevated	607657	55	100	99	87
CTNS	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750	606272	71	100	100	100
CTSA	Galactosialidosis, 256540	613111	83	100	100	98
CTSC	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650	602365	46	100	98	79
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	79	100	100	100
CTSK	Pycnodysostosis, 265800	601105	45	100	100	93
CUBN	Megaloblastic anemia-1, Finnish type, 261100	602997	59	100	98	84
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	82	100	100	100
CYP7A1	No OMIM phenotype	118455	52	100	100	94
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, 270800	603711	56	100	100	94
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	82	100	100	100
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025	614984	54	100	99	91
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	53	100	94	71
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	61	100	100	94
DLST	No OMIM phenotype	126063	61	100	100	96

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DMGDH	Dimethylglycine dehydrogenase deficiency, 605850	605849	53	100	99	89
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	606060	50	100	94	76
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	129	100	100	97
DOLK	Congenital disorder of glycosylation, type Im, 610768	610746	94	100	100	100
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	54	100	100	95
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	54	93	89	77
DPM2	Congenital disorder of glycosylation, type Iu, 615042	603564	54	100	100	93
DPM3	Congenital disorder of glycosylation, type Io, 612937	605951	132	100	100	100
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	46	100	99	85
DPYS	Dihydropyrimidinuria, 222748	613326	55	100	99	90
EBP	Chondrodysplasia punctata dominant, 302960 MEND syndrome, 300960	300205	66	100	100	96
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277	602292	51	100	99	87
EGF	Hypomagnesemia 4, renal, 611718	131530	53	100	98	84
ENO3	?Glycogen storage disease XIII, 612932	131370	95	100	100	100
EOGT	Adams-Oliver syndrome 4, 615297	614789	40	100	97	73
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	45	89	85	77
ETFA	Glutaric acidemia IIA, 231680	608053	41	100	97	78
ETFB	Glutaric acidemia IIB, 231680	130410	68	100	100	100
ETFDH	Glutaric acidemia IIC, 231680	231675	67	100	100	95
ETHE1	Ethylmalonic encephalopathy, 602473	608451	71	100	100	92
EXT1	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700	608177	51	100	97	89
EXT2	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	68	100	99	89
FAH	Tyrosinemia, type I, 276700	613871	59	100	100	93
FASTKD2	?Mitochondrial complex IV deficiency, 220110	612322	55	100	99	90
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700	611570	82	100	100	100
FECH	Protoporphyria, erythropoietic, 177000	612386	44	100	98	83

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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	49	100	98	81
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	57	98	93	88
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	99	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	65	100	100	97
FMO3	Trimethylaminuria, 602079	136132	51	100	98	90
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	85	100	100	100
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	613622	64	100	100	100
FTCD	Glutamate formiminotransferase deficiency, 229100	606806	54	97	93	89
FUCA1	Fucosidosis, 230000	612280	54	100	100	87
FXYD2	Hypomagnesemia 2, renal, 154020	601814	62	100	100	100
G6PC	Glycogen storage disease Ia, 232200	613742	64	100	100	95
GAA	Glycogen storage disease II, 232300	606800	74	100	100	99
GALC	Krabbe disease, 245200	606890	38	100	94	64
GALE	Galactose epimerase deficiency, 230350	606953	69	100	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	93	100	100	100
GALNS	Mucopolysaccharidosis IVA, 253000	612222	59	100	100	94
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 211900	601756	56	100	100	91
GALT	Galactosemia, 230400	606999	96	100	100	100

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GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	60	100	97	90
GANAB	Polycystic kidney disease 3, 600666	104160	56	100	99	92
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	44	100	100	89
GBA	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600	606463	145	100	100	100
GBE1	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	607839	49	100	95	75
GCDH	Glutaricaciduria, type I, 231670	608801	79	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	45	100	99	87
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446	606857	57	100	99	86
GCSH	Glycine encephalopathy, 605899	238330	70	100	91	57
GFM1	Combined oxidative phosphorylation deficiency 1, 609060	606639	52	100	99	88
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, 610542	138292	41	100	97	73
GGT1	No OMIM phenotype	612346	41	73	63	57
GIF	Intrinsic factor deficiency, 261000	609342	60	100	100	93
GK	Glycerol kinase deficiency, 307030	300474	47	100	91	74
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	85	100	100	100
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	67	100	99	87
GLDC	Glycine encephalopathy, 605899	238300	50	100	94	82
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	64	100	93	85
GLUL	Glutamine deficiency, congenital, 610015	138290	62	100	100	98
GLYCTK	D-glyceric aciduria, 220120	610516	108	100	100	100
GM2A	GM2-gangliosidosis, AB variant, 272750	613109	72	100	100	100
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510	615495	69	100	100	100
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352	615320	105	100	100	100

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GNE	Nonaka myopathy, 605820 Sialuria, 269921	603824	65	100	100	92
GNMT	Glycine N-methyltransferase deficiency, 606664	606628	67	100	100	98
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	59	100	98	90
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	47	100	97	86
GNPTG	Mucolipidosis III gamma, 252605	607838	103	100	98	89
GNS	Mucopolysaccharidosis type IIID, 252940	607664	46	100	98	88
GPHN	Molybdenum cofactor deficiency C, 615501	603930	48	100	99	84
GRHPR	Hyperoxaluria, primary, type II, 260000	604296	60	100	100	94
GSS	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900	601002	58	100	100	95
GSTZ1	Tyrosinemia, type Ib	603758	58	100	100	98
GUSB	Mucopolysaccharidosis VII, 253220	611499	74	100	95	85
GYG1	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199	603942	44	100	100	92
GYS1	Glycogen storage disease 0, muscle, 611556	138570	78	100	100	98
GYS2	Glycogen storage disease 0, liver, 240600	138571	46	100	99	88
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	47	100	100	92
HADHA	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015	600890	71	100	100	89
HADHB	Trifunctional protein deficiency, 609015	143450	51	100	98	87
HAL	[Histidinemia], 235800	609457	57	100	99	92
HAMP	Hemochromatosis, type 2B, 613313	606464	72	100	100	100
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	54	100	99	87
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	69	100	100	94
HFE	{Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200 [Transferrin serum level QTL2], 614193	613609	71	100	100	98
HGD	Alkaptonuria, 203500	607474	45	100	97	77

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HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	48	94	94	85
HIBADH	No OMIM phenotype	608475	36	96	88	65
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	610690	38	100	91	61
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	63	100	99	91
HMBS	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000	609806	62	100	100	99
HMGCL	HMG-CoA lyase deficiency, 246450	613898	57	100	100	94
HMGCS2	HMG-CoA synthase-2 deficiency, 605911	600234	57	100	100	93
HOGA1	Hyperoxaluria, primary, type III, 613616	613597	87	100	100	100
HPD	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	609695	72	100	100	91
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	39	100	95	75
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	70	100	100	99
HSD17B10	HDS10 mitochondrial disease, 300438	300256	63	100	100	95
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	49	100	97	80
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765	607764	90	100	100	100
HYAL1	?Mucopolysaccharidosis type IX, 601492	607071	76	100	100	100
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	60	100	100	96
IDS	Mucopolysaccharidosis II, 309900	300823	63	100	96	85
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	72	100	98	88
ISCU	Myopathy with lactic acidosis, hereditary, 255125	611911	56	100	100	90
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	58	100	99	84
ITPA	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850	147520	76	100	100	98
IVD	Isovaleric acidemia, 243500	607036	86	100	100	98
KHK	[Fructosuria], 229800	614058	75	100	100	93
KYNU	?Hydroxykynureninuria, 236800	605197	57	100	100	92
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	47	100	99	89
LAMP2	Danon disease, 300257	309060	33	100	92	58
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840	603590	63	100	100	94

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LBR	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471	600024	48	100	97	84
LCAT	Fish-eye disease, 136120 Norum disease, 245900	606967	106	100	100	100
LCT	Lactase deficiency, congenital, 223000	603202	84	100	100	98
LDHA	Glycogen storage disease XI, 612933	150000	50	100	96	72
LDLR	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890	606945	117	100	100	100
LFNG	?Spondylocostal dysostosis 3, 609813	602576	63	84	82	79
LIPA	Cholesteryl ester storage disease, 278000 Wolman disease, 278000	613497	46	100	99	82
LIPC	{Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797	151670	54	100	98	85
LIPI	{Hypertriglyceridemia, susceptibility to}, 145750	609252	48	100	98	80
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380	612625	45	100	92	67
LPL	Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11] Lipoprotein lipase deficiency, 238600	609708	74	100	100	95
LRP2	Donnai-Barrow syndrome, 222448	600073	47	100	97	84
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	44	100	95	74
MAN1B1	Mental retardation 15, 614202	604346	90	100	100	98
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	75	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	59	100	98	87
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	41	100	96	80
MAT1A	Hypermethioninemia, persistent, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, 250850	610550	65	100	100	99
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	609014	46	100	99	86
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120	608419	55	100	100	80
MCOLN1	Mucolipidosis IV, 252650	605248	84	100	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	50	100	98	81
MGAT2	Congenital disorder of glycosylation, type IIa, 212066	602616	80	100	100	100
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360	606761	56	100	97	87
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100	607481	59	100	100	97

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MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110	607568	50	100	99	84
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	76	100	100	100
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	51	100	98	78
MOCOS	Xanthinuria, type II, 603592	613274	62	100	98	84
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	69	100	100	98
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	50	100	100	90
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	88	100	100	99
MPI	Congenital disorder of glycosylation, type Ib, 602579	154550	103	100	100	99
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	137960	65	100	100	99
MRPS16	Combined oxidative phosphorylation deficiency 2, 610498	609204	58	100	100	98
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719	605810	61	100	99	84
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834	607545	81	100	100	92
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	65	100	100	98
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	52	100	98	85
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	602568	61	100	98	92
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	49	100	99	89
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	56	100	98	86
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	58	100	100	91
NAGA	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241	104170	72	100	100	100
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	80	100	97	90
NAGS	N-acetylglutamate synthase deficiency, 237310	608300	57	100	100	99
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	53	100	99	88
NDUFA1	Mitochondrial complex I deficiency, 252010 dominant	300078	83	100	100	100

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NDUFA11	Mitochondrial complex I deficiency, 252010 dominant	612638	72	100	100	100
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602137	87	100	100	100
NDUFAF1	Mitochondrial complex I deficiency, 252010 dominant	606934	49	100	99	86
NDUFAF2	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	609653	63	100	90	77
NDUFAF3	Mitochondrial complex I deficiency, 252010 dominant	612911	79	100	100	100
NDUFAF4	Mitochondrial complex I deficiency, 252010 dominant	611776	72	100	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010 dominant	612360	54	100	100	89
NDUFS1	Mitochondrial complex I deficiency, 252010 dominant	157655	49	100	98	77
NDUFS2	Mitochondrial complex I deficiency, 252010 dominant	602985	59	100	100	97
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	603846	73	100	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	602694	56	100	100	90
NDUFS6	Mitochondrial complex I deficiency, 252010 dominant	603848	52	100	100	96
NDUFS7	Leigh syndrome, 256000	601825	64	100	100	97
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000	602141	91	100	100	100
NDUFV1	Mitochondrial complex I deficiency, 252010 dominant	161015	95	100	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010 dominant	600532	47	100	96	78
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	99	100	100	100
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	93	100	100	100
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	58	100	100	93
NPC2	Niemann-pick disease, type C2, 607625	601015	64	100	100	100
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	55	100	96	80
NT5C	No OMIM phenotype	191720	70	100	85	72
NUBPL	Mitochondrial complex I deficiency, 252010 dominant	613621	53	100	100	89
NUS1	?Congenital disorder of glycosylation, type 1aa, 617082	610463	59	100	100	96
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	53	100	100	89
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	36	100	92	67
OGDH	Alpha-ketoglutarate dehydrogenase deficiency, 203740	613022	76	100	99	96

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OPA1	Behr syndrome, 210000 {Glaucoma, normal tension, susceptibility to}, 606657 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250	605290	46	100	96	77
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	66	100	100	100
OPLAH	5-oxoprolinase deficiency, 260005	614243	77	100	99	97
OTC	Ornithine transcarbamylase deficiency, 311250	300461	46	100	98	78
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	601424	50	100	99	83
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	45	100	97	82
PC	Pyruvate carboxylase deficiency, 266150	608786	90	100	100	100
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070	126090	48	100	94	94
PCCA	Propionicacidemia, 606054	232000	50	100	99	85
PCCB	Propionicacidemia, 606054	232050	53	100	96	86
PCK1	?Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency, 261680	614168	76	100	100	100
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	42	99	93	79
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	44	100	98	82
PDHX	Lacticacidemia due to PDX1 deficiency, 245349	608769	55	100	100	96
PDK1	No OMIM phenotype	602524	51	100	98	86
PDK2	No OMIM phenotype	602525	72	100	100	98
PDK3	?Charcot-Marie-Tooth disease dominant, 6, 300905	300906	39	100	90	60
PDK4	No OMIM phenotype	602527	41	100	97	75
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	72	100	100	100
PDP2	No OMIM phenotype	615499	99	100	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	50	100	93	84
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	48	100	95	78
PEPD	Prolidase deficiency, 170100	613230	58	100	98	91
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	49	100	98	84
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	54	100	100	86
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	76	100	100	98

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PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	43	100	100	87
PFKM	Glycogen storage disease VII, 232800	610681	61	100	100	94
PGAM2	Glycogen storage disease X, 261670	612931	93	100	100	100
PGAP1	Mental retardation 42, 615802	611655	51	100	95	78
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207	615187	94	100	100	99
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	62	100	100	98
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	48	100	100	91
PGM1	Congenital disorder of glycosylation, type It, 614921	171900	50	100	99	89
PGM3	Immunodeficiency 23, 615816	172100	59	100	99	89
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	103	100	100	100
PHKA1	Muscle glycogenosis, 300559	311870	39	100	96	67
PHKA2	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000	300798	42	100	95	81
PHKB	Phosphorylase kinase deficiency of liver and muscle, 261750	172490	54	100	99	91
PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027	172471	83	100	100	100
PHYH	Refsum disease, 266500	602026	80	100	92	78
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	65	100	100	99
PIGC	No OMIM phenotype	601730	118	100	100	100
PIGG	Mental retardation 53, 616917	616918	75	100	100	99
PIGL	CHIME syndrome, 280000	605947	62	100	100	81
PIGM	Glycosylphosphatidylinositol deficiency, 610293	610273	58	100	100	99
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	45	100	93	72
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	83	100	100	99
PIGQ	No OMIM phenotype	605754	96	100	100	100
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	88	100	100	100
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	71	100	100	94
PIGW	?Hyperphosphatasia with mental retardation syndrome 5, 616025	610275	74	100	100	100
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809	610662	40	100	100	100
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	53	100	100	91
PNLIP	?Pancreatic lipase deficiency, 614338	246600	42	100	98	80
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	54	100	97	89
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	53	100	100	85
POFUT1	Dowling-Degos disease 2, 615327	607491	64	100	100	99

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POGLUT1	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, type 2Z, 617232	615618	39	100	98	82
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	71	100	100	99
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions 4, 610131	604983	68	100	99	91
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123	606822	63	100	100	95
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	69	100	100	98
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	52	100	99	89
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	124015	84	100	100	100
PPOX	Porphyria variegata, 176200	600923	73	100	100	98
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	51	100	100	87
PRKAG2	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200	602743	86	100	100	93
PRKCSH	Polycystic liver disease 1, 174050	177060	72	100	100	99
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	60	100	92	81
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	43	100	96	79
PSAP	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900	176801	77	100	97	92

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PSAT1	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	610936	50	100	100	91
PSPH	Phosphoserine phosphatase deficiency, 614023	172480	37	100	90	61
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640	612719	54	100	100	79
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	608109	74	100	99	92
PYCR1	Cutis laxa, type IIB, 612940 Cutis laxa, type IIIB, 614438	179035	66	100	100	95
PYGL	Glycogen storage disease VI, 232700	613741	57	100	98	82
PYGM	McArdle disease, 232600	608455	75	100	100	100
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630	612676	52	100	97	84
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	47	100	97	76
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	71	100	100	100
RFT1	Congenital disorder of glycosylation, type In, 612015	611908	43	100	96	79
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	73	100	100	99
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	48	100	97	81
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	139	100	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	68	100	100	84
RPIA	?Ribose 5-phosphate isomerase deficiency, 608611	180430	52	100	98	89
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions 5, 613077	604712	60	100	100	95
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	49	100	93	69
SAR1B	Chylomicron retention disease, 246700	607690	53	100	100	82
SARDH	[Sarcosinemia], 268900	604455	67	100	100	97
SC5D	Lathosterolosis, 607330	602286	68	100	100	99
SCO1	Mitochondrial complex IV deficiency, 220110	603644	61	100	100	92
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	81	100	100	100
SCP2	?Leukoencephalopathy with dystonia and motor neuropathy, 613724	184755	40	100	98	78
SDHA	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165	600857	93	100	93	83
SDHAF1	Mitochondrial complex II deficiency, 252011	612848	51	100	100	100

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SDHB	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300	185470	80	100	100	100
SDHC	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373	602413	113	100	100	100
SDHD	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300	602690	111	100	100	100
SEC23B	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100	610512	52	100	96	84
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	44	100	92	66
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	90	100	96	94
SI	Sucrase-isomaltase deficiency, congenital, 222900	609845	45	100	97	80
SLC12A3	Gitelman syndrome, 263800	600968	69	100	100	98
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	60	100	99	83
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270	603941	50	100	99	89
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	54	100	100	94
SLC1A1	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232	133550	64	100	99	90
SLC1A3	Episodic ataxia, type 6, 612656	600111	66	100	100	97
SLC22A5	Carnitine deficiency, systemic primary, 212140	603377	72	100	100	97
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182	190315	66	100	100	100
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949	603667	61	100	100	91
SLC25A13	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814	603859	72	100	94	81
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	85	100	100	94
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	70	100	100	97
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138	613698	42	100	98	80

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SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	81	100	100	100
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773	600370	73	100	100	97
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950	610819	83	100	100	100
SLC25A4	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions 2, 609283	103220	74	100	100	100
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	94	100	100	100
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi-Bickel syndrome, 227810	138160	47	100	99	86
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	49	100	99	82
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	55	100	96	76
SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	69	100	100	97
SLC39A4	Acrodermatitis enteropathica, 201100	607059	100	100	100	0
SLC40A1	Hemochromatosis, type 4, 606069	604653	47	100	100	89
SLC6A19	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608893	73	100	100	98
SLC7A9	Cystinuria, 220100	604144	62	100	100	96
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	87	100	99	97
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	63	100	99	92
SSR4	Congenital disorder of glycosylation, type Iy, 300934	300090	53	100	100	86
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	40	94	84	61
STT3A	?Congenital disorder of glycosylation, type Iw, 615596	601134	43	100	99	89
STT3B	?Congenital disorder of glycosylation, type Ix, 615597	608605	64	100	99	90
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	61	100	99	93
SUGCT	Glutaric aciduria III, 231690	609187	49	100	98	82
SUMF1	Multiple sulfatase deficiency, 272200	607939	60	100	98	81
SUOX	Sulfite oxidase deficiency, 272300	606887	106	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	59	93	87	87
TACO1	Mitochondrial complex IV deficiency, 220110	612958	60	100	100	99

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TALDO1	Transaldolase deficiency, 606003	602063	86	100	100	97
TAT	Tyrosinemia, type II, 276600	613018	48	100	100	91
TAZ	Barth syndrome, 302060	300394	59	100	91	90
TCN1	No OMIM phenotype	189905	47	100	98	89
TCN2	Transcobalamin II deficiency, 275350	613441	76	100	100	99
TDO2	No OMIM phenotype	191070	37	100	95	71
TFR2	Hemochromatosis, type 3, 604250	604720	69	100	100	97
TH	Segawa syndrome, recessive, 605407	191290	66	100	100	97
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	102	100	100	100
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 617069	188250	46	100	99	86
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	73	100	100	97
TMEM199	Congenital disorder of glycosylation, type IIp, 616829	616815	54	100	100	96
TMEM5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	58	100	98	78
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	612418	78	100	100	94
TPMT	{Thiopurines, poor metabolism of, 1}, 610460	187680	58	100	87	68
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	70	100	100	99
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356	614138	44	100	95	77
TREH	Trehalase deficiency, 612119	275360	74	100	100	99
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	116	100	100	100
TRIM37	Mulibrey nanism, 253250	605073	44	100	97	80
TRMU	{Deafness, modifier of}, 580000 Liver failure, transient infantile, 613070	610230	67	100	99	89
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	55	100	97	89
TSFM	Combined oxidative phosphorylation deficiency 3, 610505	604723	60	100	100	100
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	39	100	89	62
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	45	100	96	79
TUFM	Combined oxidative phosphorylation deficiency 4, 610678	602389	100	100	100	100
TUSC3	Mental retardation 7, 611093	601385	55	100	99	88
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	109	100	100	99
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	131222	67	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	88	100	100	100
UMOD	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860	191845	62	100	99	85
UMPS	Orotic aciduria, 258900	613891	56	100	100	92
UPB1	Beta-ureidopropionase deficiency, 613161	606673	65	100	100	96
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158	191330	61	100	100	95
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159	612080	86	100	100	100
UROC1	?Urocanase deficiency, 276880	613012	59	100	100	97
UROD	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100	613521	70	100	100	98
UROS	Porphyria, congenital erythropoietic, 263700	606938	45	100	99	81
USF1	{Hyperlipidemia, familial combined, susceptibility to}, 602491	191523	81	100	100	100
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700	608547	68	100	100	96
VPS13B	Cohen syndrome, 216550	607817	54	100	98	87
XDH	Xanthinuria, type I, 278300	607633	53	100	98	85
XYLT1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	74	98	92	88
XYLT2	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822	608125	79	100	100	96
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	610957	78	100	100	96

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "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 96 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x