METABOLIC DISORDERS GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype and OMIM ID
AASS	100.0	100%	98%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	64.9	98%	90%	GABA-transaminase deficiency, 613163
ABCD1	58.2	73%	73%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, 300100
ABCD4	100.8	100%	99%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	117.1	98%	94%	Sitosterolemia, 210250
ABCG8	90.3	96%	95%	Sitosterolemia, 210250 Gallbladder disease 4, 611465
ABHD12	60.9	100%	87%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 614857
ABHD5	116.3	100%	95%	Chanarin-Dorman syndrome, 275630
ACACA	93.4	99%	97%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	103.7	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	90.8	100%	100%	ACAD9 deficiency, 611126
ACADM	144.9	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	101.2	100%	98%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	78.8	98%	95%	2-methylbutyrylglycinuria, 610006
ACADVL	88.3	100%	95%	VLCAD deficiency, 201475
ACAT1	106.8	100%	98%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	84.6	100%	98%	?ACAT2 deficiency, 614055
ACO2	77.5	89%	82%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	77.7	97%	94%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	82.9	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	129.4	100%	98%	Mental retardation, X-linked 63, 300387
ACY1	85.1	100%	96%	Aminoacylase 1 deficiency, 609924
ADA	72.3	100%	96%	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700
ADCY5	83.9	99%	94%	Dyskinesia, familial, with facial myokymia, 606703
ADK	113.5	94%	94%	Hypermethioninemia due to adenosine kinase deficiency, 614300

ADSL	126.2	100%	99%	Adenylosuccinase deficiency, 103050
AGA	111.6	100%	91%	Aspartylglucosaminuria, 208400
AGK	104.1	100%	100%	Hyperoxaluria, primary, type 1, 259900
AGL	137.7	100%	100%	Sengers syndrome, 212350 Cataract, autosomal recessive congenital 5, 614691
AGPAT2	65.7	92%	88%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPS	104.3	100%	100%	Lipodystrophy, congenital generalized, type 1, 608594
AGXT	84.9	98%	92%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AHCY	76.9	91%	69%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	91.4	100%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	70.8	78%	74%	Reticular dysgenesis, 267500
AKR1D1	96.2	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	91.9	96%	90%	Porphyria, acute hepatic, 612740 Lead poisoning, susceptibility to, 612740
ALAS2	84.4	92%	88%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	97.7	99%	95%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	83.5	94%	93%	Microphthalmia, isolated 8, 615113
ALDH2	85.1	98%	93%	Alcohol sensitivity, acute, 610251 Hangover, susceptibility to, 610251 Sublingual nitroglycerin, susceptibility to poor response to Esophageal cancer, alcohol-related, susceptibility to
ALDH3A2	92.1	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	70.7	93%	90%	Hyperprolinemia, type II, 239510
ALDH5A1	61.9	97%	90%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	96.6	100%	99%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	63.3	91%	86%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	105.2	98%	94%	Glycogen storage disease XII, 611881
ALDOB	117.9	100%	99%	Fructose intolerance, 229600
ALG1	45.6	45%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	111.8	99%	96%	Acquired long QT syndrome, reduced susceptibility to, 613688
ALG11	140.7	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	93.4	100%	97%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	113.0	96%	95%	Congenital disorder of glycosylation, type Is, 300884

ALG2	103.1	99%	89%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	81.3	98%	89%	Congenital disorder of glycosylation, type Id, 601110
ALG6	92.5	100%	99%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	84.7	97%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	80.7	99%	98%	Congenital disorder of glycosylation, type Il, 608776
ALOX12B	96.0	100%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	81.6	100%	97%	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300Hypophosphatasia, adult, 146300
AMACR	80.9	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	67.1	92%	84%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD3	81.8	99%	96%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	127.9	100%	100%	Glycine encephalopathy, 605899
AP1S1	65.2	99%	88%	MEDNIK syndrome, 609313
APOC2	168.8	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APRT	49.4	100%	94%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	121.9	98%	90%	Argininemia, 207800
ARSA	78.2	97%	94%	Metachromatic leukodystrophy, 250100
ARSB	90.7	100%	97%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	93.5	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	72.9	97%	93%	Argininosuccinic aciduria, 207900
ASPA	96.0	100%	100%	Canavan disease, 271900
ASS1	41.5	86%	54%	Citrullinemia, 215700
ATIC	109.3	100%	97%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP6V0A2	97.9	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	122.6	100%	100%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	122.5	99%	97%	Wilson disease, 277900

ATP8B1	104.8	98%	97%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480
AUH	111.3	100%	100%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	121.2	100%	100%	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	85.4	99%	97%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11), 615181
B3GALTL	108.3	100%	96%	Peters-plus syndrome, 261540
B3GAT3	55.2	91%	82%	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600
B3GNT2	121.5	100%	100%	No OMIM phenotype
B4GALT1	80.7	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	77.9	100%	96%	Ehlers-Danlos syndrome, progeroid form, 130070
BAAT	121.0	99%	96%	Hypercholanemia, familial, 607748
BCKDHA	100.9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	97.7	99%	85%	Maple syrup urine disease, type Ib, 248600
BLVRA	87.9	100%	100%	Hyperbiliverdinemia, 614156
BMP2	116.2	100%	99%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200
BPGM	140.6	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	132.0	100%	100%	Biotinidase deficiency, 253260
C1GALT1C1	164.6	100%	100%	Tn polyagglutination syndrome, somatic, 300622
CANT1	101.7	100%	97%	[Glutaric aciduria III], 231690
CAT	83.6	99%	91%	Desbuquois dysplasia, 251450
CBS	73.5	99%	81%	Acatalasemia, 614097
CEL	56.4	62%	59%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CERKL	127.5	100%	99%	Maturity-onset diabetes of the young, type VIII, 609812
CERS3	81.1	100%	99%	Ichtyosis, congenital, autosomal recessive 9, 615023

CFTR	113.1	95%	93%	Congenital bilateral absence of vas deference, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF {Bronchiestasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic},
CHIT1	72.8	90%	80%	[Chitotriosidase deficiency], 614122
CHKB	77.0	91%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	118.4	100%	97%	Ehlers-Danlos syndrome, musculocantractural type 1, 601776
CHST3	63.9	100%	98%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	123.3	100%	100%	Macular corneal dystrophy, 217800
CHSY1	142.4	96%	94%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	84.1	100%	98%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	125.0	97%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	72.1	98%	85%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	124.6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	97.2	100%	98%	No OMIM phenotype
COG1	120.4	99%	97%	Congenital disorder of glycosylation, type 2g, 611209
COG4	85.3	97%	95%	Congenital disorder of glycosylation, type 2j, 613189
COG5	99.2	100%	96%	Congenital disorder of glycosylation, type 2i, 613612
COG6	94.9	100%	96%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	80.3	99%	94%	Congenital disorder of glycosylation, type 2e, 608779
COG8	108.6	100%	100%	Congenital disorder of glycosylation, type 2h, 611182
COMT	83.0	100%	98%	Schizophrenia, susceptibility to, 181500 Panic disorder, susceptibility to, 167870
CP	85.2	98%	93%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290

CPOX	79.2	100%	98%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	101.6	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 Pulmonary hypertension, familial persistent, of the newborn, 265380 Venoocclusive disease after bone marrow transplantation
CPT1A	96.5	100%	98%	CPT deficiency, hepatic, type IA, 255120
CPT2	94.0	95%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 Encephalopathy, acute, infection-induced, 4, susceptibility to, 614212
CTH	117.2	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTNS	109.2	93%	86%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800 (3)
CTSA	97.0	100%	99%	Galactosialidosis, 256540
CTSC	94.7	100%	99%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CTSD	93.6	100%	97%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	121.0	100%	98%	Pycnodysostosis, 265800
CUBN	83.2	98%	94%	Megaloblastic anemia-1, Finnish type, 261100
CYB5R3	81.6	97%	90%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	91.5	100%	99%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	137.4	97%	93%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	101.3	98%	93%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Low renin hypertension, susceptibility to Aldosterone to renin ratio raised

CYP17A1	102.8	100%	98%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	125.2	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	108.4	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	7.4	21%	14%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	106.5	100%	96%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	96.1	100%	95%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	102.6	97%	95%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	104.8	98%	93%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	91.6	99%	94%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	61.0	98%	86%	D-2-hydroxyglutaric aciduria, 600721
DAO	99.4	100%	97%	Schizophrenia, 181500
DBH	100.7	99%	96%	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360
DBT	104.9	100%	100%	Maple syrup urine disease, type II, 248600
DCXR	89.5	100%	98%	Pentosuria, 260800
DDC	90.3	100%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	136.3	99%	93%	Spastic paraplegia 28, autosomal recessive
DDOST	103.2	100%	98%	Congenital disorder of glycosylation, type Ir, 614507
DGAT1	80.6	90%	85%	diacylglycerol o-acyltransferase 1
DGKE	110.2	98%	97%	Nephrotic syndrome, type 7, 615008
DGUOK	99.6	100%	99%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	81.2	99%	97%	Desmosterolosis, 602398
DHCR7	107.0	99%	98%	Smith-Lemli-Opitz syndrome, 270400
DHFR	50.6	79%	63%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	93.2	100%	97%	Miller syndrome, 263750
DLD	131.2	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	106.0	98%	96%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC19	55.5	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNM1L	92.9	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388

DNM2	78.1	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482
DNMT1	102.1	99%	96%	Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	93.3	100%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	142.4	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	87.5	100%	94%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	145.4	100%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	79.4	98%	98%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100.1	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPYD	112.6	99%	97%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	59.1	100%	98%	Dihydropyrimidinuria, 222748
EBP	95.6	99%	95%	Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	64.1	99%	91%	No OMIM phenotype Leigh disease (Peters (2014) Brain 137,2903)
ELOVL4	107.3	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3)
ENO3	100.3	99%	94%	Glycogen storage disease XIII, 612932
EPHX1	92.1	95%	84%	?Fetal hydantoin syndrome Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 Preeclampsia, susceptibility to, 189800
EPHX2	86.2	95%	91%	Hypercholesterolemia, familial, due to LDLR defect, modifier of, 143890
ETFA	108.5	100%	100%	Glutaric acidemia IIA, 231680
ETFB	105.7	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	120.2	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	57.6	100%	98%	Ethylmalonic encephalopathy, 602473
EXT1	103.7	97%	95%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300 (3)
EXT2	105.1	100%	95%	Exostoses, multiple, type 2, 133701
FA2H	55.7	93%	73%	Spastic paraplegia 35, autosomal recessive, 612319

FAH	111.8	100%	100%	Tyrosinemia, type I, 276700
FBP1	100.5	100%	97%	Fructose-1,6-bidphosphatase deficiency, 229700
FECH	104.2	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FH	85.3	96%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	80.2	99%	98%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 613153 Muscular dystrophy-dystroglycanopathy (with or without mental retardation), 606612 Muscular dystrophy-dystroglycanopathy, 507155
FKTN	112.3	100%	99%	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 253800 Muscular dystrophy-dystroglycanopathy (without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FMO3	105.4	98%	96%	Trimethylaminuria, 602079
FOLR1	79.3	96%	89%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	57.3	87%	80%	Glutamate formiminotransferase deficiency, 229100
FUCA1	80.4	100%	99%	Fucosidosis, 230000
FUT2	196.6	100%	100%	Norwalk virus infection, resistance to [Bombay phenotype] Vitamin B12 plasma level QTL1, 612542
FUT6	74.0	84%	68%	Fucosyltransferase 6 deficiency, 613852
G6PC	139.6	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	115.8	100%	99%	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541
G6PD	100.2	95%	95%	Hemolytic anemia due to G6PD deficiency Favism, 134700 Resistance to malaria due to G6PD deficiency, 611162
GAA	97.3	100%	98%	Glycogen storage disease II, 232300
GAD1	91.4	100%	98%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	99.5	100%	98%	Krabbe disease, 245200
GALE	116.4	100%	100%	Galactose epimerase deficiency, 230350
GALK1	93.7	100%	97%	Galactokinase deficiency with cataracts, 230200
GALNS	65.5	92%	91%	Mucopolysaccharidosis IVA, 253000
GALT	113.7	100%	100%	Galactosemia, 230400
GAMT	89.1	96%	89%	GAMT deficiency, 612736
GATM	81.7	98%	89%	AGAT deficiency, 612718

GBA	65.2	65%	62%	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013 Parkinson disease, late-onset, susceptibility to, 16860
GBA2	119.1	100%	100%	Spastic paraplegia 46, autosomal recessive
GBE1	104.1	99%	94%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	78.5	92%	87%	Glutaricaciduria, type I, 231670
GCH1	86.3	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	85.3	100%	99%	MODY, type II, 125851 Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, gestational, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176
GCLC	127.1	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 Myocardial infarction, susceptibility to, 608446
GCLM	80.2	84%	83%	Myocardial infarction, susceptibility to, 608446
GCSH	14.5	52%	37%	Glycine encephalopathy, 605899
GFPT1	99.5	99%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GK	48.0	89%	79%	Glycerol kinase deficiency, 307030
GLA	92.4	100%	98%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	79.6	98%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	57.6	97%	85%	Glycine encephalopathy, 605899
GLRA1	112.6	100%	99%	Hyperekplexia, hereditary 1, autosomal dominant or recessive,149400
GLRX5	39.1	81%	62%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	110.4	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	30.2	70%	55%	Glutamine deficiency, congenital, 610015
GLYCTK	82.9	100%	97%	D-glyceric aciduria, 220120
GM2A	125.2	100%	100%	GM2-gangliosidosis, AB variant, 272750

GMPPB	118.4	100%	100%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (with mental retardation), type B, 14, 615351
GMPS	107.3	100%	97%	Leukemia, acute myelogenous, 601626
GNE	99.1	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GNMT	85.6	100%	99%	Glycine N-methyltransferase deficiency, 606664
GNPAT	122.0	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	129.1	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	91.0	82%	80%	Mucolipidosis III gamma, 252605
GNS	80.1	97%	88%	Mucopolysaccharidosis type IIID, 252940
GOT1	104.1	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GPD1	72.9	100%	97%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100.8	100%	100%	Brugada syndrome 2, 611777
GPHN	119.9	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GPI	91.2	100%	97%	Hemolytic anemia, nonspherocytic, 613470
GPX1	12.3	73%	36%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	90.4	91%	85%	Hyperoxaluria, primary, type II, 260000
GSS	83.5	98%	97%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GUSB	67.7	89%	80%	Mucopolysaccharidosis VII, 253220
GYG1	45.8	77%	54%	Glycogen storage disease XV, 613507
GYS1	67.4	96%	77%	Glycogen storage disease 0, muscle, 611556
GYS2	90.3	100%	100%	Glycogen storage disease, type 0, 240600
H6PD	120.6	100%	100%	Cortisone reductase deficiency 1, 604931
HADH	84.4	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	92.9	93%	89%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	86.6	100%	100%	Trifunctional protein deficiency, 609015
HAGH	79.4	99%	95%	[Glyoxalase II deficiency], 614033

HEXA	91.9	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	105.5	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	93.1	100%	94%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferrin serum level QTL2], 614193
HGD	85.6	100%	100%	Alkaptonuria, 203500
HGSNAT	99.2	93%	93%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	129.1	100%	94%	No OMIM phenotype
HIBCH	61.5	100%	98%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	106.9	100%	98%	Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	140.4	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	88.3	100%	96%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	102.4	100%	98%	HMG-CoA lyase deficiency, 246450
HMGCS2	112.5	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	64.0	100%	93%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HOGA1	71.3	97%	93%	Hyperoxaluria, primary, type III, 613616
HPD	101.1	100%	98%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350 (3)
HPRT1	96.2	100%	98%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HS6ST1	6.3	24%	1%	Hypogonadotropic hypogonadism 15 with or without anosmia, 614880
HSD11B1	107.1	100%	98%	Cortisone reductase deficiency 2, 614662
HSD11B2	110.6	77%	75%	Apparent mineralocorticoid excess, 218030
HSD17B10	105.3	99%	92%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705

HSD17B3	97.4	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	91.8	100%	98%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	41.0	88%	72%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	71.9	86%	80%	Bile acid synthesis defect, congenital, 1, 607765
HYAL1	87.5	99%	96%	Mucopolysaccharidosis type IX, 601492
IDH2	107.5	100%	99%	D-2-hydrosyglutaric aciduria 2, 613657
IDH3B	124.0	95%	95%	Retinitis pigmentosa 46, 612572
IDS	101.7	90%	86%	Mucopolysaccharidosis II, 309900
IDUA	81.0	95%	89%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IMPAD1	124.9	100%	100%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	36.7	73%	61%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
INPP5E	68.4	98%	94%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300 (3)
INPPL1	92.4	97%	95%	Opsismodysplasia, 258480
ISPD	75.3	96%	91%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
IVD	96.3	100%	98%	Isovaleric acidemia, 243500
KMT2A	132.6	98%	98%	Wiedemann-Steiner syndrome, 605130 Leukemia, myeloid/lymphoid or mixed-lineage
KMT2D	102.0	99%	98%	Kabuki syndrome 1, 147920
L2HGDH	78.2	100%	96%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	109.4	100%	99%	Danon disease, 300257
LARGE	100.5	97%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LCAT	105.0	94%	88%	Norum disease, 245900 Fish-eye disease, 136120
LCT	128.5	99%	98%	Lactase deficiency, congenital, 223000
LDHA	38.1	65%	57%	Glycogen storage disease XI, 612933
LDHB	76.8	100%	100%	Lactate dehydrogenase-B deficiency, 614128

LFNG	56.0	80%	75%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LIPA	112.8	100%	100%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	89.0	97%	94%	[High density lipoprotein cholesterol level QTL 12], 612797 Diabetes mellitus, noninsulin-dependent, 125853 Hepatic lipase deficiency, 614025
LMBRD1	111.9	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	101.0	100%	97%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	75.8	99%	95%	Majeed syndrome, 609628
LPL	110.7	100%	99%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]
LRAT	187.7	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	58.8	83%	70%	Leukotriene C4 synthase deficiency, 614037
LYST	116.3	99%	97%	Muscular dystrophy, congenital, megaconial type, 602541 (3)
MAN1B1	96.6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	81.3	99%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	87.6	100%	99%	Mannosidosis, beta, 248510
MAOA	101.9	100%	100%	Brunner syndrome, 300615
MAT1A	90.0	100%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	94.6	100%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	105.2	95%	89%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	85.8	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	101.3	97%	93%	Mucolipidosis IV, 252650
MFSD8	105.4	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	194.6	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	151.3	100%	100%	Thyroid carcinoma, follicular, 188470
MLYCD	69.5	89%	84%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	160.5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	75.3	99%	92%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110

MMACHC	175.4	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	68.9	100%	100%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MOCS1	71.4	99%	93%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	112.6	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MOGS	126.1	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	117.6	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPI	96.7	100%	95%	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	73.9	100%	97%	No OMIM phenotype
MTHFD1	93.7	100%	96%	Spina bifida, folate-sensitive, susceptibility to, 601634 Abruptio placentae, susceptibility to
MTHFR	97.7	100%	97%	Homocystinuria due to MTHFR deficiency, 236250 Schizophrenia, susceptibility to, 181500 Vascular disease, susceptibility to Neural tube defects, susceptibility to, 601634 Thromboembolism, susceptibility to, 188050
MTM1	103.6	100%	100%	Myotubular myopathy, X-linked, 310400
MTMR2	96.5	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	100.3	99%	98%	Methylcobalamin deficiency, cblG type, 250940 Neural tube defects, folate-sensitive, susceptibility to, 601634
MTRR	102.8	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 Neural tube defects, folate-sensitive, susceptibility to, 601634
MUT	115.5	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MVK	86.1	100%	99%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
NAGA	82.1	100%	95%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241 (3)
NAGLU	67.0	94%	84%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	56.5	94%	76%	N-acetylglutamate synthase deficiency, 237310
NEU1	17.2	61%	35%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	93.1	100%	100%	Leber congenital amaurosis 9, 608553

NNT	95.1	100%	100%	Glucocorticoid deficiency 4, 614736
NPC1	88.3	100%	96%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	68.6	100%	100%	Niemann-pick disease, type C2, 607625
NSD1	119.7	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650 (3)
NSDHL	112.0	99%	97%	CHILD syndrome, 308050 CK syndrome, 300831 (3)
NT5C3A	67.8	95%	89%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	107.3	100%	99%	Calcification of joints and arteries, 211800
OAT	42.2	82%	62%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	112.6	98%	97%	Lowe syndrome, 309000 Dent disease 2, 300555
OPA3	107.3	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	97.5	99%	97%	5-oxoprolinase deficiency, 260005
OTC	103.3	100%	99%	Ornithine transcarbamylase deficiency, 311250
OXCT1	89.9	100%	99%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	82.4	100%	95%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	113.2	99%	93%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PC	94.0	97%	91%	Pyruvate carboxylase deficiency, 266150
PCBD1	56.7	100%	90%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	95.3	98%	95%	Propionicacidemia, 606054
PCCB	106.8	100%	100%	Propionicacidemia, 606054
PEPD	63.3	100%	89%	Prolidase deficiency, 170100
PEX1	118.6	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73.1	89%	85%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	167.9	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	114.0	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510

PEX13	134.7	98%	96%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885 (3)
PEX14	90.9	100%	99%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	86.0	92%	83%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	107.5	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	150.3	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867 (3)
PEX26	116.8	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873 (3)
PEX3	130.7	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	91.1	98%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 (3)
PEX6	89.8	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	98.2	99%	89%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	98.7	100%	98%	Glycogen storage disease VII, 232800
PGAM2	101.7	100%	100%	Glycogen storage disease X, 261670
PGAP2	125.9	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	76.4	85%	77%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	103.5	99%	96%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHGDH	85.1	100%	99%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	88.2	95%	94%	Muscle glycogenosis, 300559
PHKA2	97.0	100%	98%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHYH	85.5	100%	98%	Refsum disease, 266500
PIGA	134.6	100%	99%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGL	92.4	100%	99%	CHIME syndrome, 280000
PIGM	108.4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	102.9	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	104.9	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749

PIGT	129.9	100%	100%	Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	165.0	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	112.1	93%	91%	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Keratosis,
PIK3R1	144.8	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	76.5	90%	83%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	79.5	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	132.7	100%	99%	Corneal fleck dystrophy, 121850
PIP5K1C	64.0	88%	84%	Lethal congenital contractural syndrome 3, 611369
PKLR	121.9	100%	97%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PLA2G5	102.8	100%	100%	Fleck retina, familial benign, 228980
PLA2G6	77.8	100%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLA2G7	112.1	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278 Asthma, susceptibility to, 600807 Atopy, susceptibility to, 147050
PLCB1	107.8	99%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	89.0	100%	97%	Auriculocondylar syndrome 2, 614669
PLCD1	105.5	99%	95%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	118.0	98%	96%	Nephrotic syndrome, type 3, 610725
PLCG2	105.4	100%	99%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLIN1	44.2	92%	67%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	80.5	100%	97%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	108.2	100%	100%	Bruck syndrome 2, 609220
PLOD3	82.8	99%	88%	Lysyl hydroxylase 3 deficiency, 612394
PMM2	85.6	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	89.6	100%	99%	Pancreatic lipase deficiency, 614338

PNMT	42.3	99%	88%	?Hypertension, essential, 145500
PNP	117.2	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	76.4	97%	93%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	82.6	99%	96%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	69.7	100%	90%	Pyridoxamine 5-phosphate oxidase deficiency, 610090
POLR3A	85.6	99%	95%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100.3	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	97.4	100%	98%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 253280 Muscular dystrophy-dystroglycanopathy (with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), 613157
POMGNT2	142.2	100%	100%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 253280 Muscular dystrophy-dystroglycanopathy (with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMK	142.0	100%	100%	?Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,12, 616094 Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 615249
POMT1	94.7	100%	97%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 236670 Muscular dystrophy-dystroglycanopathy (with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle),613157
POMT2	69.5	98%	92%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 613150 Muscular dystrophy-dystroglycanopathy (with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), 613157
PPM1K	109.9	100%	92%	Maple syrup urine disease, mild variant, 615135
PPOX	92.4	100%	97%	Porphyria variegata, 176200
PPT1	63.0	100%	90%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRODH	52.0	78%	63%	Hyperprolinemia, type I, 239500 Schizophrenia, susceptibility to, 4, 600850
PRPS1	131.2	100%	100%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500

PSAP	82.2	100%	99%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSAT1	38.9	69%	49%	Phosphoserine aminotransferase deficiency, 610992
PSPH	44.6	69%	49%	Phosphoserine phosphatase deficiency, 614023
PTEN	122.4	99%	94%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 Meningioma, 607174 Glioma susceptibility 2, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome
PTGIS	57.0	98%	89%	Hypertension, essential, 145500
PTPN11	41.7	83%	68%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTS	103.0	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	81.9	100%	98%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	96.3	99%	98%	Glycogen storage disease VI, 232700
PYGM	95.8	100%	99%	McArdle disease, 232600
QDPR	75.9	100%	96%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RDH12	66.3	93%	88%	Leber congenital amaurosis 13, 612712
RDH5	99.0	100%	96%	Fundus albipunctatus, 136880
RFT1	73.9	100%	96%	Congenital disorder of glycosylation, type In, 612015
RPE65	113.6	99%	97%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	66.9	100%	99%	Ribose 5-phosphate isomerase deficiency, 608611
SARDH	74.1	98%	95%	[Sarcosinemia], 268900
SAT1	127.1	100%	100%	Keratosis follicularis spinulosa decalvans, 308800
SC5D	146.4	100%	100%	Lathosterolosis, 607330
SCARB2	96.9	100%	98%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900

SCP2	91.9	100%	97%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SEPSECS	96.3	100%	99%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	88.8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	74.9	94%	93%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SI	105.0	100%	99%	Sucrase-isomaltase deficiency, congenital, 222900
SLC16A1	142.9	100%	100%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3)
SLC17A5	95.4	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A5	108.5	100%	98%	Carnitine deficiency, systemic primary, 212140
SLC25A1	69.1	84%	79%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	92.1	100%	98%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814 (3)
SLC25A15	102.9	95%	80%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	72.3	100%	99%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4, 613710
SLC25A20	73.5	100%	96%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A38	69.5	100%	95%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC2A1	88.3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 Epilepsy, idiopathic generalized, suscpetibility to, 12, 614847 Dystonia 9, 601042
SLC2A2	115.5	100%	100%	{Diabetes mellitus, noninsulin-dependent}, 135853 Fanconi-Bickel syndrome, 227810
SLC30A10	131.8	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	96.9	100%	99%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	103.6	100%	99%	Congenital disorder of glycosylation, type 2f, 603585
SLC35C1	96.6	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	84.9	100%	97%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	69.9	100%	97%	Acrodermatitis enteropathica, 201100
SLC3A1	124.5	100%	100%	Cystinuria, 220100

SLC46A1	80.3	100%	98%	Folate malabsorption, hereditary, 229050
SLC52A1	127.6	100%	100%	Riboflavin deficiency, 615026
SLC52A2	110.6	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	73.8	99%	96%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	98.2	100%	98%	Glucose/galactose malabsorption, 606824
SLC5A2	73.7	98%	94%	Renal glucosuria, 233100
SLC6A8	7.8	20%	11%	Creatine deficiency syndrome, X-linked, 300352
SLC7A7	95.0	100%	99%	Lysinuric protein intolerance, 222700
SLC7A9	70.0	100%	100%	Cystinuria, 220100
SLCO1B1	107.7	100%	97%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	110.5	100%	95%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	103.9	97%	90%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	25.5	84%	60%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	79.1	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SPR	56.2	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	79.2	94%	88%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	102.2	100%	98%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	57.0	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	122.5	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST3GAL3	120.2	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	96.3	91%	89%	Amish infantile epilepsy syndrome, 609056
STAR	106.9	100%	100%	Lipoid adrenal hyperplasia, 201710
STS	140.6	100%	99%	Ichthyosis, X-linked, 308100
SUCLA2	78.4	94%	84%	Mitochondrial DNA depletion syndrome 5 (with methylmalonic aciduria), 612073
SUCLG1	89.4	95%	87%	Mitochondrial DNA depletion syndrome 9 (with methylmalonic aciduria), 245400
SUCLG2	76.3	98%	91%	No OMIM phenotype

SUGCT	87.6	100%	95%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
SUMF1	64.8	98%	88%	Multiple sulfatase deficiency, 272200
SUOX	176.2	100%	100%	Sulfite oxidase deficiency, 272300
TALDO1	95.3	100%	100%	Transaldolase deficiency, 606003
TAT	92.0	100%	98%	Tyrosinemia, type II, 276600
TAZ	102.3	100%	100%	Barth syndrome, 302060
TBXAS1	94.6	100%	98%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	74.8	92%	84%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	104.1	100%	97%	Transcobalamin II deficiency, 275350
TECR	83.1	100%	91%	Mental retardation, autosomal recessive 14, 614020
TH	77.8	95%	87%	Segawa syndrome, recessive, 605407
TK2	82.9	100%	95%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM165	86.6	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM5	141.3	100%	100%	Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), 615041
TMLHE	68.4	80%	78%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TPI1	63.7	96%	94%	Hemolytic anemia due to triosephosphate isomerase deficiency
TPMT	93.7	100%	100%	6-mercaptopurine sensitivity, 610460
TPP1	119.2	100%	96%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREH	102.2	91%	89%	Trehalase deficiency, 612119
TUSC3	116.9	100%	100%	Mental retardation, autosomal recessive 7, 611093
TYMP	79.2	100%	93%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	138.0	74%	74%	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 Melanoma, cutaneous malignant, suscept
TYRP1	107.1	100%	99%	Albinism, oculocutaneous, type III, 203290 Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair), 612271

UGT1A1	137.1	100%	96%	Crigler-Najjar syndrome, type I, 218800 [Gilbert syndrome], 143500 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transcient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816
UMPS	106.7	100%	100%	Orotic aciduria, 258900
UPB1	114.7	100%	97%	Beta-ureidopropionase deficiency, 613161
UROC1	72.7	99%	95%	Urocanase deficiency, 276880
UROD	84.4	99%	91%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	79.0	95%	82%	Porphyria, congenital erythropoietic, 263700
XDH	90.0	100%	99%	Xanthinuria, type I, 278300

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors