METABOLIC DISORDERS GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AASS	126.8	99%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	82.5	100%	100%	GABA-transaminase deficiency, 613163
ABCD1	107.3	80%	80%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, 300100
ABCD4	123.6	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	133.3	100%	100%	Sitosterolemia, 210250
ABCG8	136.1	98%	98%	Gallbladder disease 4, 611465 Sitosterolemia, 210250
ABHD12	100.3	98%	98%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 614857
ABHD5	224.7	100%	100%	Chanarin-Dorman syndrome, 275630
ACACA	132.4	97%	97%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	127.3	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	131.7	99%	99%	ACAD9 deficiency, 611126
ACADM	103.6	95%	95%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	123.2	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	128.1	98%	98%	2-methylbutyrylglycinuria, 610006
ACADVL	108.6	100%	100%	VLCAD deficiency, 201475
ACAT1	123.9	99%	99%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	176.9	100%	100%	?ACAT2 deficiency, 614055
ACO2	111.7	94%	94%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	157.1	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	129.6	99%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	137	97%	97%	Mental retardation, X-linked 63, 300387
ACY1	132	100%	100%	Aminoacylase 1 deficiency, 609924
ADA	93.1	97%	97%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700

ADCY5	119.1	91%	91%	Dyskinesia, familial, with facial myokymia, 606703
ADK	92.3	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	183.5	100%	100%	Adenylosuccinase deficiency, 103050
AGA	131.5	100%	100%	Aspartylglucosaminuria, 208400
AGK	119.3	97%	97%	Hyperoxaluria, primary, type 1, 259900
AGL	144.6	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	93.8	99%	99%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	49.8	92%	92%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	135.5	100%	100%	Hyperoxaluria,primary,type I,259900
AHCY	119	100%	100%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	104	98%	98%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	100.4	99%	99%	Reticular dysgenesis, 267500
AKR1D1	101	99%	99%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	97.9	100%	100%	{Lead poisoning, susceptibility to,} 612740 Porphyria, acute hepatic, 612740
ALAS2	120.4	99%	99%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	128.9	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	107	97%	97%	Microphthalmia, isolated 8, 615113
ALDH2	117	100%	100%	Alcohol sensitivity, acute, 610251 Esophageal cancer, alcohol-related, susceptibility to Hangover, susceptibility to, 610251 Sublingual nitroglycerin, susceptibility to poor response to
ALDH3A2	126.8	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	110.2	99%	99%	Hyperprolinemia, type II, 239510
ALDH5A1	76.3	91%	91%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	122.5	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	71.6	98%	98%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	171.9	95%	95%	Glycogen storage disease XII, 611881
ALDOB	147.3	100%	100%	Fructose intolerance, 229600
ALG1	53.2	53%	53%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	232.7	100%	100%	Acquired long QT syndrome, reduced susceptibility to, 613688

ALG11	155.8	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	142.9	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	122.1	100%	100%	Congenital disorder of glycosylation, type Is, 300884
ALG2	98.6	100%	100%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	114	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG6	92.1	95%	95%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	135.8	96%	96%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	118.2	100%	100%	Congenital disorder of glycosylation, type Il, 608776
ALOX12B	128.3	100%	100%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	137.7	100%	100%	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300Hypophosphatasia, adult, 146300
AMACR	135.2	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	57.6	77%	77%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD3	129.3	99%	99%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	150.4	100%	100%	Glycine encephalopathy, 605899
AP1S1	105.6	100%	100%	MEDNIK syndrome, 609313
APOC2	89.8	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APRT	61.6	100%	100%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	146.2	100%	100%	Argininemia, 207800
ARSA	90	100%	100%	Metachromatic leukodystrophy, 250100
ARSB	120.9	94%	94%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	121.3	97%	97%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	100.7	99%	99%	Argininosuccinic aciduria, 207900
ASPA	132.6	93%	93%	Canavan disease, 271900
ASS1	88.9	100%	100%	Citrullinemia, 215700
ATIC	120.1	98%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP6V0A2	136.6	100%	100%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250

ATP7A	170.7	99%	99%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	148.7	100%	100%	Wilson disease, 277900
ATP8B1	147.4	97%	97%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480
AUH	77.3	100%	100%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	103.6	100%	100%	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	117.5	92%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11), 615181
B3GALTL	102.9	99%	99%	Peters-plus syndrome, 261540
B3GAT3	88.6	99%	99%	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600
B3GNT1	108.1	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,13,615287
B4GALT1	110.2	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	103	99%	99%	Ehlers-Danlos syndrome, progeroid form, 130070
BAAT	124.3	96%	96%	Hypercholanemia, familial, 607748
BCKDHA	164.1	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	110.6	88%	88%	Maple syrup urine disease, type Ib, 248600
BCMO1	168.8	100%	100%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	133.1	100%	100%	Hyperbiliverdinemia, 614156
BMP2	168.7	99%	99%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200
BPGM	119.1	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	137.6	100%	100%	Biotinidase deficiency, 253260
C1GALT1C1	202.3	100%	100%	Tn polyagglutination syndrome, somatic, 300622
C7orf10	117	95%	95%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
CANT1	145.3	100%	100%	[Glutaric aciduria III], 231690

CAT	163.5	100%	100%	Desbuquois dysplasia, 251450
CBS	103.5	97%	97%	Acatalasemia, 614097
CEL	117.4	84%	84%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CERKL	94.2	99%	99%	Maturity-onset diabetes of the young, type VIII, 609812
CERS3	127	100%	100%	Ichtyosis, congenital, autosomal recessive 9, 615023
CFTR	121	98%	98%	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	114.9	100%	100%	[Chitotriosidase deficiency], 614122
CHKB	89.1	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	170.4	95%	95%	Ehlers-Danlos syndrome, musculocantractural type 1, 601776
CHST3	82.7	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	285.9	100%	100%	Macular corneal dystrophy, 217800
CHSY1	125.5	96%	96%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	99.3	96%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	147	97%	97%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131	98%	98%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	203.2	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	131.2	96%	96%	3-methylglutaconic aciduria,type VII,with cataracts,neurologic involvement and neutropenia,616271
COG1	120.7	100%	100%	Congenital disorder of glycosylation, type 2g, 611209
COG4	116.3	100%	100%	Congenital disorder of glycosylation, type 2j, 613189
COG5	104.6	95%	95%	Congenital disorder of glycosylation, type 2i, 613612
COG6	84.8	93%	93%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	125.6	100%	100%	Congenital disorder of glycosylation, type 2e, 608779
COG8	101.4	100%	100%	Congenital disorder of glycosylation, type 2h, 611182

COMT	173.7	100%	100%	Schizophrenia, susceptibility to, 181500 Panic disorder, susceptibility to, 167870
CP	129.5	93%	93%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPOX	106.4	95%	95%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	144.6	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 Pulmonary hypertension, familial persistent, of the newborn, 265380 Venoocclusive disease after bone marrow transplantation
CPT1A	164	100%	100%	CPT deficiency, hepatic, type IA, 255120
CPT2	145.2	98%	98%	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	168.4	98%	98%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTNS	119.2	100%	100%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800 (3)
CTSA	124	100%	100%	Galactosialidosis, 256540
CTSC	131.1	100%	100%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CTSD	153.4	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	103.8	100%	100%	Pycnodysostosis, 265800
CUBN	124.2	99%	99%	Megaloblastic anemia-1, Finnish type, 261100
CYB5R3	155	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	113.7	97%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743

CYP11B1	152.9	100%	100%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	154.4	100%	100%	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	119.5	100%	100%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	180.9	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	103.2	100%	100%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	15.7	57%	57%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	152.5	97%	97%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	115.1	100%	100%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	149.2	99%	99%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	120.2	94%	94%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	93	91%	91%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	130.7	97%	97%	D-2-hydroxyglutaric aciduria, 600721
DAO	121.8	100%	100%	Schizophrenia, 181500
DBH	133.5	100%	100%	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360
DBT	100.1	99%	99%	Maple syrup urine disease, type II, 248600
DCXR	148.5	100%	100%	Pentosuria, 260800
DDC	99.7	99%	99%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	151	96%	96%	Spastic paraplegia 28, autosomal recessive,609340
DDOST	103	100%	100%	Congenital disorder of glycosylation, type Ir, 614507
DGAT1	134.6	92%	92%	?Diarrhea 7,615863
DGKE	154.3	96%	96%	Nephrotic syndrome, type 7, 615008

DGUOK	125	96%	96%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	173.7	100%	100%	Desmosterolosis, 602398
DHCR7	139.4	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHFR	41.7	79%	79%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	91.4	100%	100%	Miller syndrome, 263750
DLD	125.1	99%	99%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	152.4	99%	99%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC19	92.3	99%	99%	3-methylglutaconic aciduria, type V, 610198
DNM1L	112.8	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	126.1	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482
DNMT1	109.5	99%	99%	Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	119.4	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	169.5	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	115.6	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	129.6	90%	90%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	97.3	100%	100%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	135.5	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPYD	156.6	94%	94%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	121.4	100%	100%	Dihydropyrimidinuria, 222748
EBP	114.8	100%	100%	Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	110.3	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency,616277
ELOVL4	104	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	168.4	99%	99%	Glycogen storage disease XIII, 612932

EPHX1	128.5	99%	99%	?Fetal hydantoin syndrome Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 Preeclampsia, susceptibility to, 189800
EPHX2	110.5	100%	100%	Hypercholesterolemia, familial, due to LDLR defect, modifier of, 143890
ETFA	137.4	100%	100%	Glutaric acidemia IIA, 231680
ETFB	100.1	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	95.7	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	74.5	100%	100%	Ethylmalonic encephalopathy, 602473
EXT1	86.2	99%	99%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	159	100%	100%	Exostoses, multiple, type 2, 133701
FA2H	99.5	94%	94%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	149.1	100%	100%	Tyrosinemia, type I, 276700
FBP1	106.1	100%	100%	Fructose-1,6-bidphosphatase deficiency, 229700
FECH	120.8	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FH	150.2	89%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	82.2	100%	100%	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, 507155
FKTN	135.4	95%	95%	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-d
FMO3	148.3	100%	100%	Trimethylaminuria, 602079
FOLR1	137	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	86	91%	91%	Glutamate formiminotransferase deficiency, 229100
FUCA1	134.1	100%	100%	Fucosidosis, 230000

FUT2	165.5	100%	100%	Norwalk virus infection, resistance to [Bombay phenotype] Vitamin B12 plasma level QTL1, 612542
FUT6	169	100%	100%	Fucosyltransferase 6 deficiency, 613852
G6PC	178.6	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	132.1	100%	100%	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541
G6PD	151.1	100%	100%	Hemolytic anemia due to G6PD deficiency Favism, 134700 Resistance to malaria due to G6PD deficiency, 611162
GAA	103.6	100%	100%	Glycogen storage disease II, 232300
GAD1	114.8	100%	100%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	98.5	95%	95%	Krabbe disease, 245200
GALE	145	100%	100%	Galactose epimerase deficiency, 230350
GALK1	101.4	97%	97%	Galactokinase deficiency with cataracts, 230200
GALNS	91.9	96%	96%	Mucopolysaccharidosis IVA, 253000
GALT	146	100%	100%	Galactosemia, 230400
GAMT	107.1	98%	98%	GAMT deficiency, 612736
GATM	165.9	100%	100%	AGAT deficiency, 612718
GBA	199.6	100%	100%	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	165.2	100%	100%	Spastic paraplegia 46, autosomal recessive
GBE1	146.2	99%	99%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	136.4	92%	92%	Glutaricaciduria, type I, 231670
GCH1	80.5	92%	92%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910

GCK	133.6	100%	100%	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	122.8	99%	99%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 Myocardial infarction, susceptibility to, 608446
GCLM	110.7	97%	97%	Myocardial infarction, susceptibility to, 608446
GCSH	30.6	63%	63%	Glycine encephalopathy, 605899
GFPT1	130.7	99%	99%	Myasthenia, congenital, with tubular aggregates 1, 610542
GK	55.3	76%	76%	Glycerol kinase deficiency, 307030
GLA	82.5	100%	100%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	81.8	100%	100%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	75.5	87%	87%	Glycine encephalopathy, 605899
GLRA1	112.6	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive,149400
GLRX5	87.2	99%	99%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	72.1	92%	92%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	92.3	100%	100%	Glutamine deficiency, congenital, 610015
GLYCTK	197.3	100%	100%	D-glyceric aciduria, 220120
GM2A	113	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPB	229.7	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), t
GMPS	103.6	98%	98%	Leukemia, acute myelogenous, 601626

GNE	158.4	100%	100%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GNMT	157.9	100%	100%	Glycine N-methyltransferase deficiency, 606664
GNPAT	145.4	97%	97%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	166.9	97%	97%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	130.1	96%	96%	Mucolipidosis III gamma, 252605
GNS	104.4	97%	97%	Mucopolysaccharidosis type IIID, 252940
GOT1	127.5	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GPD1	96.1	100%	100%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	151.1	100%	100%	Brugada syndrome 2, 611777
GPHN	164.4	96%	96%	Molybdenum cofactor deficiency, type C, 252150
GPI	133.4	100%	100%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPX1	35.2	85%	85%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	110.9	84%	84%	Hyperoxaluria, primary, type II, 260000
GSS	96.5	100%	100%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GUSB	113.9	89%	89%	Mucopolysaccharidosis VII, 253220
GYG1	134.4	100%	100%	Glycogen storage disease XV, 613507
GYS1	102.6	98%	98%	Glycogen storage disease 0, muscle, 611556
GYS2	161.9	99%	99%	Glycogen storage disease, type 0, 240600
H6PD	149.1	99%	99%	Cortisone reductase deficiency 1, 604931
HADH	109.9	98%	98%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	78.5	97%	97%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	90	98%	98%	Trifunctional protein deficiency, 609015
HAGH	116.5	98%	98%	[Glyoxalase II deficiency], 614033

HEXA	124.3	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	136.4	97%	97%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	126.4	98%	98%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferr
HGD	126.9	97%	97%	Alkaptonuria, 203500
HGSNAT	102.7	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	90.4	91%	91%	No OMIM phenotype
HIBCH	63.9	93%	93%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	141	99%	99%	Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	156.8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	109.7	100%	100%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	137.2	100%	100%	HMG-CoA lyase deficiency, 246450
HMGCS2	131.2	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	132.2	99%	99%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HOGA1	130.7	100%	100%	Hyperoxaluria, primary, type III, 613616
HPD	125.5	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	79.7	98%	98%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HS6ST1	62	95%	95%	Hypogonadotropic hypogonadism 15 with or without anosmia, 614880
HSD11B1	170.1	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	154.8	85%	85%	Apparent mineralocorticoid excess, 218030
HSD17B10	132.9	100%	100%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B3	144.5	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300

HSD17B4	91.9	93%	93%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	146.3	100%	100%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	124.5	99%	99%	Bile acid synthesis defect, congenital, 1, 607765
HYAL1	112.5	100%	100%	Mucopolysaccharidosis type IX, 601492
IDH2	93	100%	100%	D-2-hydrosyglutaric aciduria 2, 613657
IDH3B	165.1	100%	100%	Retinitis pigmentosa 46, 612572
IDS	131.9	100%	100%	Mucopolysaccharidosis II, 309900
IDUA	104.1	92%	92%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IMPAD1	126	100%	100%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	50	90%	90%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
INPP5E	90.9	96%	96%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300 (3)
INPPL1	110.2	99%	99%	Opsismodysplasia, 258480
ISPD	106.2	95%	95%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
IVD	112.4	100%	100%	Isovaleric acidemia, 243500
KMT2A	148.9	98%	98%	Wiedemann-Steiner syndrome, 605130 Leukemia, myeloid/lymphoid or mixed-lineage
KMT2D	135.4	100%	100%	Kabuki syndrome 1, 147920
L2HGDH	116.7	97%	97%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	141.8	92%	92%	Danon disease, 300257
LARGE	136.2	100%	100%	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	136.5	100%	100%	Norum disease, 245900 Fish-eye disease, 136120
LCT	118.5	99%	99%	Lactase deficiency, congenital, 223000
LDHA	59	92%	92%	Glycogen storage disease XI, 612933
LDHB	102.1	88%	88%	Lactate dehydrogenase-B deficiency, 614128

LFNG	87.9	84%	84%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LIPA	113.9	96%	96%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	115.8	100%	100%	[High density lipoprotein cholesterol level QTL 12], 612797 Diabetes mellitus, noninsulin-dependent, 125853 Hepatic lipase deficiency, 614025
LMBRD1	72.5	92%	92%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	126.5	99%	99%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	106	100%	100%	Majeed syndrome, 609628
LPL	158.6	100%	100%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]
LRAT	249.7	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	48.8	69%	69%	Leukotriene C4 synthase deficiency, 614037
LYST	137	97%	97%	Chediak-Higashi syndrome,214500
MAN1B1	130.1	100%	100%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	121.9	97%	97%	Mannosidosis, alpha-, types I and II, 248500
MANBA	127.4	100%	100%	Mannosidosis, beta, 248510
MAOA	167	100%	100%	Brunner syndrome, 300615
MAT1A	168.9	97%	97%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	146.6	100%	100%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	129.4	100%	100%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	105	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	141.2	98%	98%	Mucolipidosis IV, 252650
MFSD8	127.8	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	160.2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	150.2	99%	99%	Thyroid carcinoma, follicular, 188470
MLYCD	77.4	92%	92%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	162.6	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100

MMAB	114.1	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	171.8	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	66.8	85%	85%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MOCS1	69.2	94%	94%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	131.9	99%	99%	Molybdenum cofactor deficiency, type B, 252150
MOGS	107.7	99%	99%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	114.1	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPI	115	100%	100%	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	38.4	87%	87%	Microcephaly,congenital cataract and psoriasiform dermatitis,616834
MTHFD1	136.4	99%	99%	Spina bifida, folate-sensitive, susceptibility to, 601634 Abruptio placentae, susceptibility to
MTHFR	142.1	100%	100%	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	128.9	100%	100%	Myotubular myopathy, X-linked, 310400
MTMR2	110.3	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	139.6	99%	99%	Methylcobalamin deficiency, cblG type, 250940 Neural tube defects, folate-sensitive, susceptibility to, 601634
MTRR	119.2	100%	100%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 Neural tube defects, folate-sensitive, susceptibility to, 601634
MUT	109.8	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MVK	123.3	100%	100%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
NAGA	129.9	100%	100%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241 (3)
NAGLU	99.6	93%	93%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920

NAGS	66.9	97%	97%	N-acetylglutamate synthase deficiency, 237310
NEU1	16	66%	66%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	115.2	99%	99%	Leber congenital amaurosis 9, 608553
NNT	140.9	99%	99%	Glucocorticoid deficiency 4, 614736
NPC1	142.7	100%	100%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	118	100%	100%	Niemann-pick disease, type C2, 607625
NSD1	146.6	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650
NSDHL	218.9	100%	100%	CHILD syndrome, 308050 CK syndrome, 300831 (3)
NT5C3A	61.7	87%	87%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	160.1	100%	100%	Calcification of joints and arteries, 211800
OAT	78.1	76%	76%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	166.9	99%	99%	Lowe syndrome, 309000 Dent disease 2, 300555
OPA3	101.1	95%	95%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	94.5	99%	99%	5-oxoprolinase deficiency, 260005
OTC	169	100%	100%	Ornithine transcarbamylase deficiency, 311250
OXCT1	109.6	97%	97%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	168.9	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	151.2	100%	100%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PC	139.5	100%	100%	Pyruvate carboxylase deficiency, 266150
PCBD1	97.7	99%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	101.3	91%	91%	Propionicacidemia, 606054
PCCB	140.7	95%	95%	Propionicacidemia, 606054
PEPD	107.1	100%	100%	Prolidase deficiency, 170100
PEX1	103.4	97%	97%	Peroxisome biogenesis disorder 1A (Zellweger), 214100

				Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	100.4	99%	99%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	102.3	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	137.8	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	169.7	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885 (3)
PEX14	141.3	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	114.8	92%	92%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	103.5	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	142.7	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867 (3)
PEX26	71.6	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873 (3)
PEX3	103.4	97%	97%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	105.9	100%	100%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 (3)
PEX6	68.9	89%	89%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	130.3	91%	91%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	146.6	100%	100%	Glycogen storage disease VII, 232800
PGAM2	141.8	100%	100%	Glycogen storage disease X, 261670
PGAP2	159.9	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	69.1	90%	90%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	124.7	100%	100%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHGDH	121.9	100%	100%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	150.6	99%	99%	Muscle glycogenosis, 300559
PHKA2	140.5	100%	100%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHYH	68	100%	100%	Refsum disease, 266500

PIGA	104.1	94%	94%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGL	126	100%	100%	CHIME syndrome, 280000
PIGM	142.4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	109	93%	93%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	115.2	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	144	100%	100%	Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	144.8	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	117.9	100%	100%	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	121.5	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	79.9	89%	89%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	98.6	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	137.9	100%	100%	Corneal fleck dystrophy, 121850
PIP5K1C	101.1	96%	96%	Lethal congenital contractural syndrome 3, 611369
PKLR	156.2	100%	100%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PLA2G5	125.4	100%	100%	Fleck retina, familial benign, 228980
PLA2G6	106.6	99%	99%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLA2G7	120.4	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278 Asthma, susceptibility to, 600807 Atopy, susceptibility to, 147050
PLCB1	153.4	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	129.1	99%	99%	Auriculocondylar syndrome 2, 614669
PLCD1	96.5	100%	100%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600

PLCE1	149.8	99%	99%	Nephrotic syndrome, type 3, 610725
PLCG2	116.5	100%	100%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLIN1	72.4	99%	99%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	127.5	100%	100%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	108.9	89%	89%	Bruck syndrome 2, 609220
PLOD3	102.5	98%	98%	Lysyl hydroxylase 3 deficiency, 612394
PMM2	143	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	169.2	100%	100%	Pancreatic lipase deficiency, 614338
PNMT	77.2	98%	98%	?Hypertension, essential, 145500
PNP	126.7	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	116	100%	100%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	118.5	99%	99%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	71.3	100%	100%	Pyridoxamine 5-phosphate oxidase deficiency, 610090
POLR3A	137.2	100%	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	148.1	99%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	108.2	100%	100%	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),
POMGNT2	241.6	100%	100%	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), typ
POMK	206.7	100%	100%	?Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,12, 615249

POMT1	146	100%	100%	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),
POMT2	104	98%	98%	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),
PPM1K	147.7	100%	100%	Maple syrup urine disease, mild variant, 615135
PPOX	89.3	100%	100%	Porphyria variegata, 176200
PPT1	175.7	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRODH	79.2	92%	92%	Hyperprolinemia, type I, 239500 Schizophrenia, susceptibility to, 4, 600850
PRPS1	216.4	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	109.8	100%	100%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSAT1	47.9	83%	83%	Phosphoserine aminotransferase deficiency, 610992
PSPH	134.3	92%	92%	Phosphoserine phosphatase deficiency, 614023
PTEN	141.6	100%	100%	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	113.6	95%	95%	Hypertension, essential, 145500

PTPN11	86.4	98%	98%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTS	121.3	94%	94%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	88.7	99%	99%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	163.7	100%	100%	Glycogen storage disease VI, 232700
PYGM	136.6	100%	100%	McArdle disease, 232600
QDPR	75.8	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RDH12	87.2	95%	95%	Leber congenital amaurosis 13, 612712
RDH5	137.7	100%	100%	Fundus albipunctatus, 136880
RFT1	96.6	99%	99%	Congenital disorder of glycosylation, type In, 612015
RPE65	135.1	100%	100%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	128.8	94%	94%	Ribose 5-phosphate isomerase deficiency, 608611
SARDH	114.4	91%	91%	[Sarcosinemia], 268900
SAT1	186.7	100%	100%	Keratosis follicularis spinulosa decalvans, 308800
SC5D	214.2	100%	100%	Lathosterolosis, 607330
SCARB2	112.9	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	108.6	100%	100%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SEPSECS	154.9	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	110.4	99%	99%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	122.2	97%	97%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SI	92.1	96%	96%	Sucrase-isomaltase deficiency, congenital, 222900
SLC16A1	134.8	99%	99%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3)
SLC17A5	103.1	97%	97%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A5	152.3	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC25A1	72.7	97%	97%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182

SLC25A13	111.3	98%	98%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814 (3)
SLC25A15	192.7	97%	97%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	70.8	100%	100%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	102.6	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A38	109	100%	100%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC2A1	159.4	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 Epilepsy, idiopathic generalized, suscpetibility to, 12, 614847 Dystonia 9, 601042
SLC2A2	166.4	100%	100%	{Diabetes mellitus, noninsulin-dependent}, 135853 Fanconi-Bickel syndrome, 227810
SLC30A10	168	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	128.7	95%	95%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	131.2	100%	100%	Congenital disorder of glycosylation, type 2f, 603585
SLC35C1	191.6	98%	98%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	113.4	100%	100%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	74.7	99%	99%	Acrodermatitis enteropathica, 201100
SLC3A1	176.3	100%	100%	Cystinuria, 220100
SLC46A1	92	99%	99%	Folate malabsorption, hereditary, 229050
SLC52A1	188.2	100%	100%	Riboflavin deficiency, 615026
SLC52A2	146.9	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	108.6	100%	100%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	132.9	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	127.9	100%	100%	Renal glucosuria, 233100
SLC6A8	67.3	93%	93%	Creatine deficiency syndrome, X-linked, 300352
SLC7A7	105.7	100%	100%	Lysinuric protein intolerance, 222700

SLC7A9	134.6	100%	100%	Cystinuria, 220100
SLCO1B1	49	91%	91%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	51.8	90%	90%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	112.8	99%	99%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	79.6	94%	94%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	168.5	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SPR	154.6	99%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	115.6	97%	97%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	149.4	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	72	100%	100%	Pseudovaginal perineoscrotal hypospadias,264600
SRD5A3	145.7	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST3GAL3	158.4	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	122.9	94%	94%	Amish infantile epilepsy syndrome, 609056
STAR	116.5	100%	100%	Lipoid adrenal hyperplasia, 201710
STS	124.7	99%	99%	Ichthyosis, X-linked, 308100
SUCLA2	61.5	92%	92%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with methylmalonic aciduria), 612073
SUCLG1	105.3	100%	100%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	52.8	90%	90%	No OMIM phenotype
SUMF1	109.5	94%	94%	Multiple sulfatase deficiency, 272200
SUOX	198	100%	100%	Sulfite oxidase deficiency, 272300
TALDO1	132.9	100%	100%	Transaldolase deficiency, 606003
TANGO2	129.7	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TAT	123.7	100%	100%	Tyrosinemia, type II, 276600
TAZ	127.2	100%	100%	Barth syndrome, 302060
TBXAS1	130.1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	107.9	93%	93%	Osteopetrosis, autosomal recessive 1, 259700

TCN2	157.4	100%	100%	Transcobalamin II deficiency, 275350
TECR	78.1	98%	98%	Mental retardation, autosomal recessive 14, 614020
TH	67.9	96%	96%	Segawa syndrome, recessive, 605407
TK2	102	91%	91%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM165	111.2	99%	99%	Congenital disorder of glycosylation, type IIk, 614727
TMEM5	122.6	91%	91%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMLHE	125.5	100%	100%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TPI1	104.6	100%	100%	Hemolytic anemia due to triosephosphate isomerase deficiency
TPMT	40.6	86%	86%	6-mercaptopurine sensitivity, 610460
TPP1	130.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREH	124.4	100%	100%	Trehalase deficiency, 612119
TUSC3	137.7	100%	100%	Mental retardation, autosomal recessive 7, 611093
TYMP	97.3	96%	96%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	159.3	100%	100%	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	181.4	100%	100%	Albinism, oculocutaneous, type III, 203290 Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair), 612271
UGT1A1	204.3	100%	100%	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	179	100%	100%	Orotic aciduria, 258900
UPB1	151.5	100%	100%	Beta-ureidopropionase deficiency, 613161
UROC1	132	100%	100%	Urocanase deficiency, 276880
UROD	167.2	100%	100%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	110.6	100%	100%	Porphyria, congenital erythropoietic, 263700
XDH	105.4	100%	100%	Xanthinuria, type I, 278300

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
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
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors