METABOLIC DISORDERS GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM ID*
AASS	101.1	100%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	64.3	97%	89%	GABA-transaminase deficiency, 613163
ABCD1	32.6	74%	67%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, 300100
ABCD4	102.8	100%	97%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	135.1	99%	87%	Sitosterolemia, 210250
ABCG8	90.7	97%	96%	Sitosterolemia, 210250 Gallbladder disease 4, 611465
ABHD12	61	98%	83%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 614857
ABHD5	121.2	100%	100%	Chanarin-Dorman syndrome, 275630
ACACA	95.6	98%	96%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	87.2	97%	92%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	91	100%	99%	ACAD9 deficiency, 611126
ACADM	154.1	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	109.2	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	85.7	98%	96%	2-methylbutyrylglycinuria, 610006
ACADVL	87.7	100%	97%	VLCAD deficiency, 201475
ACAT1	108.9	100%	98%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	102.5	100%	100%	?ACAT2 deficiency, 614055
ACO2	78.6	90%	84%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	75.3	99%	93%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	77	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	66.8	100%	97%	Mental retardation, X-linked 63, 300387
ACY1	85.9	100%	97%	Aminoacylase 1 deficiency, 609924
ADA	70.7	100%	95%	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700
ADCY5	83.6	98%	94%	Dyskinesia, familial, with facial myokymia, 606703
ADK	112.6	94%	94%	Hypermethioninemia due to adenosine kinase deficiency, 614300

ADSL	124.2	100%	98%	Adenylosuccinase deficiency, 103050
AGA	117.2	100%	89%	Aspartylglucosaminuria, 208400
AGK	107.5	99%	99%	Hyperoxaluria, primary, type 1, 259900
AGL	146.4	100%	100%	Sengers syndrome, 212350 Cataract, autosomal recessive congenital 5, 614691
AGPAT2	60.2	94%	79%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPS	116.3	100%	100%	Lipodystrophy, congenital generalized, type 1, 608594
AGXT	88.5	97%	90%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AHCY	70.6	92%	73%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	82.3	100%	98%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	75.1	79%	77%	Reticular dysgenesis, 267500
AKR1D1	96.5	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	83.5	100%	95%	Porphyria, acute hepatic, 612740 Lead poisoning, susceptibility to, 612740
ALAS2	41	87%	79%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	91.9	97%	91%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	73.4	87%	82%	Microphthalmia, isolated 8, 615113
ALDH2	88.5	98%	93%	Alcohol sensitivity, acute, 610251 Hangover, susceptibility to, 610251
ALDH3A2	98.2	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	70.5	94%	88%	Hyperprolinemia, type II, 239510
ALDH5A1	64.5	97%	94%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100.9	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	71.6	94%	92%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	106.3	98%	94%	Glycogen storage disease XII, 611881
ALDOB	106.5	100%	98%	Fructose intolerance, 229600
ALG1	47.7	45%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	134.7	99%	98%	Acquired long QT syndrome, reduced susceptibility to, 613688
ALG11	154.3	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	103.4	100%	97%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	57.9	93%	84%	Congenital disorder of glycosylation, type Is, 300884
ALG2	116.5	100%	97%	Congenital disorder of glycosylation, type Ii, 607906

ALG3	87.7	100%	94%	Congenital disorder of glycosylation, type Id, 601110
ALG6	104	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	87.9	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	90.7	100%	98%	Congenital disorder of glycosylation, type Il, 608776
ALOX12B	101.5	100%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	80.4	100%	100%	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300
AMACR	94.2	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	65.8	89%	85%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD3	87.5	100%	97%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	123.5	100%	99%	Glycine encephalopathy, 605899
AP1S1	79.9	100%	99%	MEDNIK syndrome, 609313
APOC2	156.4	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APRT	45.1	96%	83%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	135	98%	91%	Argininemia, 207800
ARSA	88	97%	93%	Metachromatic leukodystrophy, 250100
ARSB	94	100%	98%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	99.1	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	80.4	99%	95%	Argininosuccinic aciduria, 207900
ASPA	120.8	100%	99%	Canavan disease, 271900
ASS1	41.2	84%	61%	Citrullinemia, 215700
ATIC	115.4	100%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP6V0A2	106.5	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	60.3	100%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	126.1	100%	98%	Wilson disease, 277900
ATP8B1	113.5	99%	98%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480

AUH	103.9	91%	90%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	125.3	100%	100%	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	83.7	91%	89%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11), 615181
B3GALTL	107.6	95%	95%	Peters-plus syndrome, 261540
B3GAT3	55.9	92%	79%	Joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600
B4GALT1	77	97%	97%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	89.7	100%	95%	Ehlers-Danlos syndrome, progeroid form, 130070
BAAT	123	100%	99%	Hypercholanemia, familial, 607748
BCKDHA	104.7	100%	98%	Maple syrup urine disease, type Ia, 248600
BCKDHB	86.1	98%	83%	Maple syrup urine disease, type Ib, 248600
BCMO1	132.5	100%	99%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	84.3	100%	99%	Hyperbiliverdinemia, 614156
BMP2	115.8	100%	100%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200
BPGM	140	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	140.1	100%	100%	Biotinidase deficiency, 253260
C1GALT1C1	87.1	100%	99%	Tn polyagglutination syndrome, somatic, 300622
C7orf10	84	91%	91%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
CANT1	91	99%	96%	[Glutaric aciduria III], 231690
CAT	91.7	98%	91%	Desbuquois dysplasia, 251450
CBS	76.2	99%	91%	Acatalasemia, 614097
CEL	57.3	64%	61%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CERKL	137.4	100%	100%	Maturity-onset diabetes of the young, type VIII, 609812
CERS3	84.7	100%	100%	Ichtyosis, congenital, autosomal recessive 9, 615023
CFTR	123.1	95%	95%	Congenital bilateral absence of vas deference, 277180 Cystic fibrosis, 219700
CHIT1	71.5	90%	84%	[Chitotriosidase deficiency], 614122
CHKB	91.1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	130.2	100%	100%	Ehlers-Danlos syndrome, musculocantractural type 1, 601776
CHST3	67.2	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095

CHST6	123.3	100%	100%	Macular corneal dystrophy, 217800
CHSY1	147.3	98%	97%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	83.5	97%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.7	100%	90%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	65	98%	82%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	133.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	101.9	96%	95%	3-methylglutaconic aciduria,type VII,with cataracts,neurologic involvement and neutropenia,616271
COG1	121	100%	98%	Congenital disorder of glycosylation, type 2g, 611209
COG4	84.4	98%	95%	Congenital disorder of glycosylation, type 2j, 613189
COG5	107.1	98%	95%	Congenital disorder of glycosylation, type 2i, 613612
COG6	101.9	97%	95%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	77	100%	96%	Congenital disorder of glycosylation, type 2e, 608779
COG8	117.1	100%	100%	Congenital disorder of glycosylation, type 2h, 611182
COMT	85.8	100%	99%	Schizophrenia, susceptibility to, 181500 Panic disorder, susceptibility to, 167870
CP	90.4	99%	92%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPOX	85.2	100%	97%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	107.7	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 Pulmonary hypertension, familial persistent, of the newborn, 265380
CPT1A	95.5	99%	97%	CPT deficiency, hepatic, type IA, 255120
CPT2	109.4	92%	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	129.7	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated

CTNS	118.7	97%	95%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTSA	96.1	100%	99%	Galactosialidosis, 256540
CTSC	88.2	100%	100%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CTSD	96.2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	129.9	100%	100%	Pycnodysostosis, 265800
CUBN	88.4	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
CYB5R3	77.9	97%	94%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	89.8	100%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	110	98%	94%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	104.9	98%	91%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400
CYP17A1	105.5	99%	96%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	135.3	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	103.3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	2.1	6%	0%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	118.1	100%	96%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	107.1	100%	98%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	107.1	99%	96%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	108.4	100%	99%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	97.1	100%	96%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	60.6	95%	85%	D-2-hydroxyglutaric aciduria, 600721
DAO	106.8	100%	100%	Schizophrenia, 181500

DBH	99	100%	97%	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360
DBT	111.8	100%	100%	Maple syrup urine disease, type II, 248600
DCXR	93.1	100%	100%	Pentosuria, 260800
DDC	89.8	100%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	138.5	100%	98%	Spastic paraplegia 28, autosomal recessive,609340
DDOST	97.6	100%	98%	Congenital disorder of glycosylation, type Ir, 614507
DGAT1	85.1	91%	88%	?Diarrhea 7,615863
DGKE	113.9	100%	99%	Nephrotic syndrome, type 7, 615008
DGUOK	96.6	100%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	88.5	99%	97%	Desmosterolosis, 602398
DHCR7	115.9	100%	97%	Smith-Lemli-Opitz syndrome, 270400
DHFR	51.9	81%	58%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	100.6	100%	99%	Miller syndrome, 263750
DLD	143.7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	118.4	97%	96%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC19	57.8	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNM1L	104.1	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	80.9	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482
DNMT1	101.7	99%	96%	Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	91.5	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	145.3	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	96.6	99%	95%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	143.3	90%	90%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	70.8	99%	93%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	101.2	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPYD	121.9	98%	96%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	60.6	100%	98%	Dihydropyrimidinuria, 222748
EBP	43.9	94%	73%	Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	64	98%	92%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency,616277

ELOVL4	102.9	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	106.8	99%	96%	Glycogen storage disease XIII, 612932
EPHX1	94	95%	91%	?Fetal hydantoin syndrome Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 Preeclampsia, susceptibility to, 189800
EPHX2	96.6	95%	93%	Hypercholesterolemia, familial, due to LDLR defect, modifier of, 143890
ETFA	117.6	100%	100%	Glutaric acidemia IIA, 231680
ETFB	107.8	100%	99%	Glutaric acidemia IIB, 231680
ETFDH	132.6	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	57	99%	88%	Ethylmalonic encephalopathy, 602473
EXT1	104.7	98%	95%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	117.4	100%	96%	Exostoses, multiple, type 2, 133701
FA2H	64.6	87%	76%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	108.6	100%	98%	Tyrosinemia, type I, 276700
FBP1	84.2	100%	95%	Fructose-1,6-bidphosphatase deficiency, 229700
FECH	106.9	100%	98%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FH	85.7	98%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	80.2	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital w/wo mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy, 507155
FKTN	108.3	100%	99%	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
FMO3	106.4	100%	100%	Trimethylaminuria, 602079
FOLR1	82.4	100%	98%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	56.5	93%	81%	Glutamate formiminotransferase deficiency, 229100
FUCA1	74.3	100%	96%	Fucosidosis, 230000
FUT2	211.3	100%	100%	Vitamin B12 plasma level QTL1, 612542
FUT6	78.2	82%	74%	Fucosyltransferase 6 deficiency, 613852

G6PC	131.2	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	116.3	100%	100%	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541
G6PD	58.3	95%	92%	Hemolytic anemia due to G6PD deficiency Favism, 134700 Resistance to malaria due to G6PD deficiency, 611162
GAA	97.7	100%	98%	Glycogen storage disease II, 232300
GAD1	96.2	100%	98%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	96.8	100%	97%	Krabbe disease, 245200
GALE	108.6	100%	100%	Galactose epimerase deficiency, 230350
GALK1	91.1	99%	98%	Galactokinase deficiency with cataracts, 230200
GALNS	71.3	93%	93%	Mucopolysaccharidosis IVA, 253000
GALT	104.8	100%	98%	Galactosemia, 230400
GAMT	103.8	98%	95%	GAMT deficiency, 612736
GATM	89.2	100%	94%	AGAT deficiency, 612718
GBA	62.5	62%	57%	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	128.7	100%	100%	Spastic paraplegia 46, autosomal recessive
GBE1	104.6	98%	94%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	76.3	93%	89%	Glutaricaciduria, type I, 231670
GCH1	109.1	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	82.5	100%	97%	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	133.6	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 Myocardial infarction, susceptibility to, 608446

GCLM	92.8	84%	84%	Myocardial infarction, susceptibility to, 608446
GCSH	14.2	48%	34%	Glycine encephalopathy, 605899
GFPT1	105.9	100%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GK	25.6	78%	57%	Glycerol kinase deficiency, 307030
GLA	47.2	95%	86%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	76.1	99%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	60.1	98%	85%	Glycine encephalopathy, 605899
GLRA1	113.9	100%	98%	Hyperekplexia, hereditary 1, autosomal dominant or recessive,149400
GLRX5	29.9	72%	46%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	111.8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	30.6	62%	52%	Glutamine deficiency, congenital, 610015
GLYCTK	83.9	99%	97%	D-glyceric aciduria, 220120
GM2A	107.6	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPB	119.9	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GMPS	116.3	100%	100%	Leukemia, acute myelogenous, 601626
GNE	98.4	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GNMT	85.8	100%	99%	Glycine N-methyltransferase deficiency, 606664
GNPAT	115.9	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	133.9	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	89.9	86%	80%	Mucolipidosis III gamma, 252605
GNS	79.1	95%	86%	Mucopolysaccharidosis type IIID, 252940
GOT1	95.9	95%	95%	Aspartate aminotransferase, serum level of, QTL1, 614419
GPD1	72.9	100%	99%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	105.6	100%	100%	Brugada syndrome 2, 611777
GPHN	119.8	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GPI	91.9	100%	97%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470

GPX1	16.2	66%	46%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	72.7	80%	64%	Hyperoxaluria, primary, type II, 260000
GSS	80	98%	94%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GUSB	61.9	89%	81%	Mucopolysaccharidosis VII, 253220
GYG1	50.3	83%	60%	Glycogen storage disease XV, 613507
GYS1	64.1	95%	81%	Glycogen storage disease 0, muscle, 611556
GYS2	92.1	100%	100%	Glycogen storage disease, type 0, 240600
H6PD	124.9	99%	99%	Cortisone reductase deficiency 1, 604931
HADH	85	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	93.2	96%	88%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	93.9	100%	99%	Trifunctional protein deficiency, 609015
HAGH	83.7	99%	97%	[Glyoxalase II deficiency], 614033
HEXA	91.1	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	114.8	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	103.5	100%	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
HGD	89.4	100%	100%	Alkaptonuria, 203500
HGSNAT	91.5	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	130.2	99%	92%	No OMIM phenotype
HIBCH	68.2	100%	99%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	111.5	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	141.8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	99.3	99%	97%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000

HMGCL	102.5	100%	99%	HMG-CoA lyase deficiency, 246450
HMGCS2	117.6	100%	99%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	65.9	100%	95%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HOGA1	64	100%	93%	Hyperoxaluria, primary, type III, 613616
HPD	98.3	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	50.8	100%	78%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HS6ST1	7.3	14%	0%	Hypogonadotropic hypogonadism 15 with or without anosmia, 614880
HSD11B1	103.1	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	113.2	78%	78%	Apparent mineralocorticoid excess, 218030
HSD17B10	54.1	95%	91%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B3	105	99%	96%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95.9	97%	96%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	44	93%	78%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	69.2	89%	76%	Bile acid synthesis defect, congenital, 1, 607765
HYAL1	94.9	99%	93%	Mucopolysaccharidosis type IX, 601492
IDH2	106.8	100%	94%	D-2-hydrosyglutaric aciduria 2, 613657
IDH3B	122.4	100%	100%	Retinitis pigmentosa 46, 612572
IDS	51	82%	72%	Mucopolysaccharidosis II, 309900
IDUA	86.7	95%	86%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IMPAD1	130.4	100%	100%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	37.9	87%	65%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
INPP5E	74.2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPPL1	94.9	98%	95%	Opsismodysplasia, 258480
ISPD	88.9	95%	94%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643

IVD	91.7	100%	95%	Isovaleric acidemia, 243500
KMT2A	142	99%	98%	Wiedemann-Steiner syndrome, 605130 Leukemia, myeloid/lymphoid or mixed-lineage
KMT2D	100.9	99%	98%	Kabuki syndrome 1, 147920
L2HGDH	79.9	94%	92%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	61.4	98%	90%	Danon disease, 300257
LARGE	105.2	99%	95%	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	107.1	93%	88%	Norum disease, 245900 Fish-eye disease, 136120
LCT	130	100%	99%	Lactase deficiency, congenital, 223000
LDHA	50.4	80%	64%	Glycogen storage disease XI, 612933
LDHB	89.5	100%	100%	Lactate dehydrogenase-B deficiency, 614128
LFNG	58.5	84%	79%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LIPA	104.1	95%	95%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	91.9	100%	95%	[High density lipoprotein cholesterol level QTL 12], 612797 Diabetes mellitus, noninsulin-dependent, 125853 Hepatic lipase deficiency, 614025
LMBRD1	108.9	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	99.2	100%	99%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	75.5	100%	97%	Majeed syndrome, 609628
LPL	109.4	100%	100%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]
LRAT	196.5	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	64	87%	66%	Leukotriene C4 synthase deficiency, 614037
LYST	120.5	99%	96%	Chediak-Higashi syndrome,214500
MAN1B1	96.6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	80.2	97%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	92.3	100%	99%	Mannosidosis, beta, 248510
MAOA	55.6	100%	95%	Brunner syndrome, 300615

MAT1A	94.5	100%	99%	Hypermethioninemia, persistent,due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	97	99%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	113.6	98%	92%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	79.6	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	96.7	97%	92%	Mucolipidosis IV, 252650
MFSD8	120	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	217.2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	149.4	100%	100%	Thyroid carcinoma, follicular, 188470
MLYCD	78.3	94%	81%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	174.5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	79.1	100%	92%	Methylmalonic aciduria, vitamin B12-responsive, 251110
MMACHC	177.9	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	72	89%	89%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MOCS1	82	99%	95%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	115.8	99%	99%	Molybdenum cofactor deficiency, type B, 252150
MOGS	126.4	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	115.5	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPI	91.2	100%	95%	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	93.3	100%	97%	No OMIM phenotype
MTHFD1	98.7	99%	95%	Spina bifida, folate-sensitive, susceptibility to, 601634 Abruptio placentae, susceptibility to
MTHFR	93	100%	98%	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	54	100%	96%	Myotubular myopathy, X-linked, 310400
MTMR2	106.9	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	106	100%	99%	Methylcobalamin deficiency, cblG type, 250940 Neural tube defects, folate-sensitive, susceptibility to, 601634

MTRR	105.2	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 Neural tube defects, folate-sensitive, susceptibility to, 601634
MUT	124.8	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MVK	88.3	100%	97%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
NAGA	78.6	100%	99%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241 (3)
NAGLU	67.1	94%	89%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	52.2	85%	75%	N-acetylglutamate synthase deficiency, 237310
NEU1	14.5	62%	29%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	93.1	100%	100%	Leber congenital amaurosis 9, 608553
NNT	96.8	100%	100%	Glucocorticoid deficiency 4, 614736
NPC1	90.6	99%	98%	Niemann-Pick disease, 257220
NPC2	61	100%	99%	Niemann-pick disease, type C2, 607625
NSD1	123.8	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650
NSDHL	48	96%	93%	CHILD syndrome, 308050 CK syndrome, 300831 (3)
NT5C3A	72.8	96%	88%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	110.3	100%	100%	Calcification of joints and arteries, 211800
OAT	44.9	83%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	58.4	97%	94%	Lowe syndrome, 309000 Dent disease 2, 300555
OPA3	108	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	97.3	99%	96%	5-oxoprolinase deficiency, 260005
OTC	54.5	100%	94%	Ornithine transcarbamylase deficiency, 311250
OXCT1	96.2	100%	98%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	83.8	96%	94%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600

PANK2	112.1	90%	86%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PC	96.3	95%	92%	Pyruvate carboxylase deficiency, 266150
PCBD1	60.2	100%	98%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	95.7	96%	94%	Propionicacidemia, 606054
PCCB	106.5	100%	98%	Propionicacidemia, 606054
PEPD	66.9	97%	89%	Prolidase deficiency, 170100
PEX1	124.5	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73	95%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	157.2	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	133	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	129.7	100%	95%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885 (3)
PEX14	92.3	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	91.8	94%	84%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	104.7	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	159.3	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867 (3)
PEX26	107.1	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873 (3)
PEX3	141.1	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	84.4	97%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 (3)
PEX6	91.8	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	93.8	90%	85%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	103.2	100%	100%	Glycogen storage disease VII, 232800
PGAM2	96.8	100%	100%	Glycogen storage disease X, 261670
PGAP2	120.3	100%	99%	Hyperphosphatasia with mental retardation syndrome 3, 614207

PGK1	42.7	75%	67%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	109.2	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHGDH	86.2	100%	97%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	48.2	96%	90%	Muscle glycogenosis, 300559
PHKA2	49.6	97%	85%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHYH	87.7	100%	99%	Refsum disease, 266500
PIGA	66.6	99%	98%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGL	97.5	100%	100%	CHIME syndrome, 280000
PIGM	105.4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	106.6	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	107.5	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	133.6	100%	99%	Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	182.7	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	126.7	93%	90%	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
PIK3R1	141.9	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	80.8	96%	87%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	72.9	100%	98%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	137.9	100%	100%	Corneal fleck dystrophy, 121850
PIP5K1C	57	85%	81%	Lethal congenital contractural syndrome 3, 611369
PKLR	123.2	100%	97%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PLA2G5	108.2	100%	100%	Fleck retina, familial benign, 228980
PLA2G6	77	99%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953

PLA2G7	122.2	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278 Asthma, susceptibility to, 600807 Atopy, susceptibility to, 147050
PLCB1	113	100%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	93.3	100%	98%	Auriculocondylar syndrome 2, 614669
PLCD1	95.1	97%	92%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	122.1	99%	97%	Nephrotic syndrome, type 3, 610725
PLCG2	107.3	99%	98%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLIN1	54.5	90%	78%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	76.1	100%	97%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	113.3	100%	100%	Bruck syndrome 2, 609220
PLOD3	79.6	95%	85%	Lysyl hydroxylase 3 deficiency, 612394
PMM2	92.4	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	86.7	100%	100%	Pancreatic lipase deficiency, 614338
PNMT	46.6	95%	92%	?Hypertension, essential, 145500
PNP	124.9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	78.5	100%	91%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	82.5	100%	97%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	71.8	100%	97%	Pyridoxamine 5-phosphate oxidase deficiency, 610090
POLR3A	90.6	99%	96%	Leukodystrophy, hypomyelinating, 7, with oligodontia and hypogonadotropic hypogonadism, 607694
POLR3B	104.2	100%	99%	Leukodystrophy, hypomyelinating, 8, with oligodontia and hypogonadotropic hypogonadism, 614381
POMGNT1	98.3	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151
POMGN2	135.3	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
POMK	128.9	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	103.9	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	74.5	100%	93%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
PPM1K	97.7	100%	95%	Maple syrup urine disease, mild variant, 615135

PPOX	99	100%	97%	Porphyria variegata, 176200
PPT1	72	100%	96%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRODH	51.5	84%	66%	Hyperprolinemia, type I, 239500 Schizophrenia, susceptibility to, 4, 600850
PRPS1	62.3	99%	97%	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	81.7	100%	98%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSAT1	45	74%	59%	Phosphoserine aminotransferase deficiency, 610992
PSPH	46.7	78%	44%	Phosphoserine phosphatase deficiency, 614023
PTEN	137.7	100%	97%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 Meningioma, 607174 Glioma susceptibility 2, 613028 Macrocephaly/autism syndrome, 605309
PTGIS	56.1	100%	89%	Hypertension, essential, 145500
PTPN11	46.9	88%	66%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTS	118.4	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	83	100%	96%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	99.8	100%	99%	Glycogen storage disease VI, 232700
PYGM	92.4	100%	98%	McArdle disease, 232600
QDPR	77.2	100%	98%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RDH12	63.6	91%	80%	Leber congenital amaurosis 13, 612712
RDH5	107.3	100%	100%	Fundus albipunctatus, 136880

RFT1	78.3	100%	97%	Congenital disorder of glycosylation, type In, 612015
RPE65	114.7	100%	98%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	72	100%	95%	Ribose 5-phosphate isomerase deficiency, 608611
SARDH	66.7	88%	85%	[Sarcosinemia], 268900
SAT1	80	100%	97%	Keratosis follicularis spinulosa decalvans, 308800
SC5D	175.4	100%	100%	Lathosterolosis, 607330
SCARB2	88.5	100%	95%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	96.3	99%	97%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SEPSECS	104.5	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	89.3	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	75.2	94%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SI	107.2	100%	99%	Sucrase-isomaltase deficiency, congenital, 222900
SLC16A1	137.9	100%	99%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3)
SLC17A5	100.9	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A5	124.2	100%	99%	Carnitine deficiency, systemic primary, 212140
SLC25A1	77.6	88%	82%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	99	100%	99%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814 (3)
SLC25A15	100.6	88%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	71.1	100%	97%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	75.1	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A38	74.4	100%	96%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC2A1	84.9	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 Epilepsy, idiopathic generalized, suscpetibility to, 12, 614847 Dystonia 9, 601042
SLC2A2	125.4	100%	100%	{Diabetes mellitus, noninsulin-dependent}, 135853 Fanconi-Bickel syndrome, 227810
SLC30A10	133.1	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280

SLC33A1	100.8	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	111.1	100%	100%	Congenital disorder of glycosylation, type 2f, 603585
SLC35C1	100.2	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	83.8	99%	97%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	65.8	100%	98%	Acrodermatitis enteropathica, 201100
SLC3A1	132	96%	96%	Cystinuria, 220100
SLC46A1	83.3	100%	97%	Folate malabsorption, hereditary, 229050
SLC52A1	132	100%	100%	Riboflavin deficiency, 615026
SLC52A2	116.3	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	69.1	100%	95%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	99.5	100%	99%	Glucose/galactose malabsorption, 606824
SLC5A2	81.4	98%	95%	Renal glucosuria, 233100
SLC6A8	4.4	13%	5%	Creatine deficiency syndrome, X-linked, 300352
SLC7A7	90.3	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	69.4	100%	96%	Cystinuria, 220100
SLCO1B1	113.3	100%	99%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	115.8	100%	98%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	110.7	99%	92%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	15.1	58%	33%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	100.5	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SPR	67.2	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	81	95%	90%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	110	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A3	130.2	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST3GAL3	113.8	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	107.6	94%	94%	Amish infantile epilepsy syndrome, 609056
STAR	100.6	100%	100%	Lipoid adrenal hyperplasia, 201710
STS	69.1	98%	92%	Ichthyosis, X-linked, 308100

SUCLA2	81.6	94%	91%	Mitochondrial DNA depletion syndrome 5, 612073
SUCLG1	94.7	95%	91%	Mitochondrial DNA depletion syndrome 9, 245400
SUCLG2	84	94%	93%	No OMIM phenotype
SUMF1	75.1	100%	97%	Multiple sulfatase deficiency, 272200
SUOX	170.4	100%	100%	Sulfite oxidase deficiency, 272300
TALDO1	91.9	100%	100%	Transaldolase deficiency, 606003
TAT	100.6	100%	100%	Tyrosinemia, type II, 276600
TAZ	47.3	100%	98%	Barth syndrome, 302060
TBXAS1	99.7	100%	99%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	76.7	95%	84%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	103.4	100%	97%	Transcobalamin II deficiency, 275350
TECR	84.1	100%	89%	Mental retardation, autosomal recessive 14, 614020
TH	84.5	96%	87%	Segawa syndrome, recessive, 605407
TK2	91.6	100%	99%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM165	92.2	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM5	168.4	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMLHE	36.6	84%	74%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TPI1	68	100%	97%	Hemolytic anemia due to triosephosphate isomerase deficiency
TPMT	95.7	100%	100%	6-mercaptopurine sensitivity, 610460
TPP1	135.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREH	105.8	95%	90%	Trehalase deficiency, 612119
TUSC3	126.2	100%	99%	Mental retardation, autosomal recessive 7, 611093
TYMP	88.5	99%	91%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	132.9	74%	74%	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952
TYRP1	125.9	100%	100%	Albinism, oculocutaneous, type III, 203290
UGT1A1	70.1	71%	61%	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	111.2	100%	100%	Orotic aciduria, 258900

UPB1	133.7	100%	100%	Beta-ureidopropionase deficiency, 613161
UROC1	76.4	98%	85%	Urocanase deficiency, 276880
UROD	84.5	97%	90%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	73.7	99%	89%	Porphyria, congenital erythropoietic, 263700
XDH	89.2	100%	99%	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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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