METABOLIC DISORDERS GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AMACR	173.1	100%	99%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	69.7	89%	76%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	151.5	100%	99%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	146.1	99%	98%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	187.9	100%	100%	Glycine encephalopathy, 605899
AP1S1	114.6	100%	99%	MEDNIK syndrome, 609313
APOC2	120.1	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APRT	73.3	100%	99%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	181	100%	100%	Argininemia, 207800
ARSA	114.5	100%	99%	Metachromatic leukodystrophy, 250100
ARSB	137.9	98%	94%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	147.1	99%	97%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	128.8	100%	98%	Argininosuccinic aciduria, 207900
ASPA	144.7	99%	94%	Canavan disease, 271900
ASS1	118.2	96%	90%	Citrullinemia, 215700
ATIC	142.5	99%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP6V0A2	160.2	99%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	148.2	99%	98%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	170.2	100%	99%	Wilson disease, 277900
ATP8B1	160.4	96%	94%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
AUH	103	99%	96%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	145.1	100%	99%	[Blood group, globoside system], 615021

				[Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	141.5	93%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALTL	120.7	98%	95%	Peters-plus syndrome, 261540
B3GAT3	105.3	98%	92%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT1	124.3	100%	99%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	122.7	97%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
B4GAT1	125.5	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	143	98%	95%	Hypercholanemia, familial, 607748
BCKDHA	195.9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	147.9	93%	84%	Maple syrup urine disease, type Ib, 248600
BCMO1	176.4	100%	100%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	155.1	100%	99%	Hyperbiliverdinemia, 614156
BMP2	203.5	100%	100%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200
BPGM	127.2	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	168.1	100%	100%	Biotinidase deficiency, 253260
C1GALT1C1	167.1	99%	98%	Tn polyagglutination syndrome, somatic, 300622
C7orf10	153.9	96%	93%	Glutaric aciduria III, 231690
CAD	174.8	99%	99%	?Congenital disorder of glycosylation, type Iz, 616457
CANT1	165.9	100%	99%	Desbuquois dysplasia 1, 251450
CAT	179.4	100%	100%	Acatalasemia, 614097
CBS	134.1	98%	94%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CEL	147	85%	83%	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	121.8	99%	96%	Retinitis pigmentosa 26, 608380
CERS3	143.1	100%	99%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	158.3	99%	97%	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic}, 167800
CHIT1	134.2	99%	98%	[Chitotriosidase deficiency], 614122

CHKB	102.6	99%	97%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	194.6	96%	94%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	107.5	99%	98%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	338.2	100%	100%	Macular corneal dystrophy, 217800
CHSY1	142.1	96%	94%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	133	99%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	155.4	99%	97%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	144.8	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	273.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	153.8	96%	96%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
AASS	169.7	100%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	103.8	100%	99%	GABA-transaminase deficiency, 613163
ABCD1	94.7	76%	69%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	152.7	99%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	167.3	100%	99%	Sitosterolemia, 210250
ABCG8	166.5	99%	97%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	113.2	98%	90%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	256.2	100%	99%	Chanarin-Dorfman syndrome, 275630
ACACA	146.4	98%	98%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	145.6	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	157.1	99%	97%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADM	135.5	99%	98%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	149.2	99%	98%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	139.7	98%	94%	2-methylbutyrylglycinuria, 610006
ACADVL	127.9	99%	97%	VLCAD deficiency, 201475
ACAT1	139	99%	97%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	193.2	100%	100%	?ACAT2 deficiency, 614055
ACO2	141.2	97%	94%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289

ACOX1	167.6	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	146.1	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	128.1	97%	93%	Mental retardation, X-linked 63, 300387
ACY1	162.7	99%	98%	Aminoacylase 1 deficiency, 609924
ADA	128.2	99%	98%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCY5	148.6	93%	91%	Dyskinesia, familial, with facial myokymia, 606703
ADK	118.9	99%	98%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	196.2	100%	100%	Adenylosuccinase deficiency, 103050
AGA	175.8	100%	100%	Aspartylglucosaminuria, 208400
AGK	152.2	99%	97%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	184.8	99%	99%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	117.9	99%	95%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	69.6	98%	91%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	168.5	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHCY	132.6	100%	98%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	141.8	99%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	110.8	99%	94%	Reticular dysgenesis, 267500
AKR1D1	131	99%	95%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	106.6	100%	98%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	99.8	98%	95%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	143.9	100%	100%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH1A3	122.3	95%	90%	Microphthalmia, isolated 8, 615113
ALDH2	137.6	100%	99%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}

ALDH3A2	153.1	100%	99%	Sjogren-Larsson syndrome, 270200
ALDH4A1	137.2	100%	99%	Hyperprolinemia, type II, 239510
ALDH5A1	105.3	92%	84%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	143.6	100%	99%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	88.5	97%	91%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	199.4	99%	96%	Glycogen storage disease XII, 611881
ALDOB	168.3	99%	98%	Fructose intolerance, 229600
ALG1	57.5	53%	49%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	355.7	100%	100%	{Long QT syndrome, acquired, reduced susceptibility to}, 613688
ALG11	189.4	100%	99%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	171.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	104.2	99%	96%	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	115.3	100%	99%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	116.7	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	126.3	98%	95%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	147.2	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	128.6	99%	99%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALOX12B	139.6	99%	98%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	164.9	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
COG1	129.4	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	139.6	100%	99%	Congenital disorder of glycosylation, type IIj, 613489
COG5	135.2	99%	95%	Congenital disorder of glycosylation, type IIi, 613612
COG6	103.3	97%	90%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	139.6	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	118.4	99%	98%	Congenital disorder of glycosylation, type IIh, 611182
COMT	248.3	100%	100%	{Panic disorder, susceptibility to}, 167870 {Schizophrenia, susceptibility to}, 181500
CP	151.7	94%	91%	Cerebellar ataxia, 604290

				Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPOX	136.6	93%	87%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	183.3	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	189.9	99%	98%	CPT deficiency, hepatic, type IA, 255120
CPT2	182	98%	96%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CTH	206.8	100%	99%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTNS	142.7	100%	99%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTSA	153.2	99%	99%	Galactosialidosis, 256540
CTSC	142.5	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	197.2	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	118.2	100%	99%	Pycnodysostosis, 265800
CUBN	134	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CYB5R3	181.8	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	142.4	99%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	187.7	100%	99%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	190.1	100%	99%	Aldosterone to renin ratio raised Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}
CYP17A1	143.7	100%	99%	17,20-lyase deficiency, isolated, 202110

				17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	200.3	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	129.2	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	15.8	56%	32%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	206.9	98%	96%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	147.5	100%	99%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	156	98%	94%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	162.9	95%	93%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	129.5	97%	92%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	153.2	98%	96%	D-2-hydroxyglutaric aciduria, 600721
DAO	142.8	100%	99%	{Schizophrenia}, 181500
DBH	173.8	100%	100%	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]
DBT	138.6	99%	96%	Maple syrup urine disease, type II, 248600
DCXR	178.5	99%	95%	[Pentosuria], 260800
DDC	121.6	99%	97%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	168.6	97%	95%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	133.1	100%	99%	?Congenital disorder of glycosylation, type Ir, 614507
DGAT1	173	91%	88%	?Diarrhea 7, 615863
DGKE	160	99%	97%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	141.9	99%	99%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	205.7	100%	100%	Desmosterolosis, 602398
DHCR7	173	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHFR	61.8	95%	81%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	102.8	100%	99%	Miller syndrome, 263750
DLD	157	99%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	188.5	99%	97%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	165.1	87%	87%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	115.5	98%	94%	3-methylglutaconic aciduria, type V, 610198

DNM1L	143.6	100%	98%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	145	99%	95%	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150
DNMT1	128.6	99%	98%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	143.6	99%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	189.5	100%	99%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	119.1	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	149.2	92%	87%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	99	99%	96%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	181.4	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPYD	190.3	97%	95%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	144.7	99%	98%	Dihydropyrimidinuria, 222748
EBP	89.2	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	123.3	100%	99%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL4	115.5	100%	99%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
ENO3	204.6	100%	100%	?Glycogen storage disease XIII, 612932
EPHX1	139.7	98%	96%	Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 ?Fetal hydantoin syndrome {Preeclampsia, susceptibility to}, 189800
EPHX2	120.9	100%	99%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
ETFA	163.2	100%	99%	Glutaric acidemia IIA, 231680
ETFB	128.4	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	127.9	100%	99%	Glutaric acidemia IIC, 231680
ETHE1	92	99%	97%	Ethylmalonic encephalopathy, 602473
EXT1	105.5	99%	97%	Chondrosarcoma, 215300

				Exostoses, multiple, type 1, 133700
EXT2	178.8	99%	99%	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
FA2H	101.9	95%	89%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	176.7	100%	99%	Tyrosinemia, type I, 276700
FBP1	132.9	100%	99%	Fructose-1,6-bisphosphatase deficiency, 229700
FECH	137.3	100%	99%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FH	183.5	93%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	103.2	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 (limb-girdle), type C, 5, 607155
FKTN	155.6	99%	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-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	200.1	99%	97%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	195.2	100%	99%	Trimethylaminuria, 602079
FOLR1	155.8	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	104.6	95%	89%	Glutamate formiminotransferase deficiency, 229100
FUCA1	151.4	100%	99%	Fucosidosis, 230000
FUT2	188.8	100%	100%	[Bombay phenotype] {Norwalk virus infection, resistance to} {Vitamin B12 plasma level QTL1}, 612542
FUT6	168.7	100%	99%	Fucosyltransferase 6 deficiency, 613852
G6PC	201.1	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	143.5	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	138.1	99%	96%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	136.8	100%	99%	Glycogen storage disease II, 232300
GAD1	139.7	99%	98%	?Cerebral palsy, spastic quadriplegic, 1, 603513

GALC	115.9	99%	96%	Krabbe disease, 245200
GALE	182.8	100%	99%	Galactose epimerase deficiency, 230350
GALK1	127.8	99%	96%	Galactokinase deficiency with cataracts, 230200
GALNS	108.4	99%	95%	Mucopolysaccharidosis IVA, 253000
GALT	182.8	100%	100%	Galactosemia, 230400
GAMT	123	98%	91%	Cerebral creatine deficiency syndrome 2, 612736
GATM	173	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GBA	227.1	100%	100%	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	185.6	100%	99%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	189.8	99%	98%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	139.2	93%	91%	Glutaricaciduria, type I, 231670
GCH1	92.5	97%	89%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	151.4	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	161.4	100%	99%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	137.2	99%	95%	{Myocardial infarction, susceptibility to}, 608446
GCSH	41.7	85%	70%	Glycine encephalopathy, 605899
GFPT1	171.9	99%	99%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GK	57.5	84%	68%	Glycerol kinase deficiency, 307030
GLA	83.5	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	97.3	99%	97%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650

				Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	88.5	92%	84%	Glycine encephalopathy, 605899
GLRA1	124.8	100%	99%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRX5	105.8	91%	84%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	84.5	94%	85%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	104.4	99%	97%	Glutamine deficiency, congenital, 610015
GLYCTK	230.9	99%	99%	D-glyceric aciduria, 220120
GM2A	154.1	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPB	276.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GMPS	141.7	98%	95%	No OMIM phenotype Leukemia, acute myelogenous, 601626
GNE	167.4	100%	99%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	176.1	99%	94%	Glycine N-methyltransferase deficiency, 606664
GNPAT	176.5	99%	97%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	190.4	99%	97%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	169.7	96%	92%	Mucolipidosis III gamma, 252605
GNS	119.6	97%	93%	Mucopolysaccharidosis type IIID, 252940
GOT1	135.7	99%	98%	Aspartate aminotransferase, serum level of, QTL1, 614419
GPD1	100.2	100%	99%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	184.1	100%	99%	Brugada syndrome 2, 611777
GPHN	186.4	99%	97%	Molybdenum cofactor deficiency C, 615501
GPI	162.7	100%	99%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPX1	37.4	89%	72%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	118	86%	81%	Hyperoxaluria, primary, type II, 260000
GSS	117	100%	99%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GUSB	126.8	91%	87%	Mucopolysaccharidosis VII, 253220
GYG1	168.1	100%	99%	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507

GYS1	120.2	99%	98%	Glycogen storage disease 0, muscle, 611556
GYS2	172	99%	97%	Glycogen storage disease 0, liver, 240600
H6PD	194.2	99%	99%	Cortisone reductase deficiency 1, 604931
HADH	126.2	97%	94%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	96.1	97%	92%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	106.7	96%	89%	Trifunctional protein deficiency, 609015
HAGH	135.4	98%	95%	[Glyoxalase II deficiency], 614033
HEXA	133.7	100%	98%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	166.2	99%	95%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	148.7	100%	99%	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HGD	144.2	99%	99%	Alkaptonuria, 203500
HGSNAT	122.6	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	120.7	93%	91%	No OMIM phenotype
HIBCH	87.8	97%	86%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	152.9	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	182.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	117.7	100%	99%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	152.8	100%	99%	HMG-CoA lyase deficiency, 246450
HMGCS2	143.1	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	152.2	96%	90%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963

HOGA1	156.5	99%	98%	Hyperoxaluria, primary, type III, 613616
HPD	159.2	100%	99%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	82.8	96%	89%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	71.3	94%	83%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD11B1	182.6	100%	99%	Cortisone reductase deficiency 2, 614662
HSD11B2	179.5	88%	84%	Apparent mineralocorticoid excess, 218030
HSD17B10	119.7	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B3	161.7	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	125.3	95%	93%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	181.3	100%	100%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	166.7	99%	96%	Bile acid synthesis defect, congenital, 1, 607765
HYAL1	104.3	100%	100%	?Mucopolysaccharidosis type IX, 601492
IDH2	108	99%	98%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	170.6	100%	100%	Retinitis pigmentosa 46, 612572
IDS	113.9	99%	97%	Mucopolysaccharidosis II, 309900
IDUA	120.5	92%	86%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IMPAD1	152.8	100%	99%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	59.3	93%	82%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
INPP5E	109	97%	92%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	142.3	98%	95%	Opsismodysplasia, 258480
ISPD	130.8	97%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
IVD	123.5	100%	99%	Isovaleric acidemia, 243500
KMT2A	165.3	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	158.6	100%	99%	Kabuki syndrome 1, 147920

L2HGDH	139.7	98%	97%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	120.4	93%	91%	Danon disease, 300257
LARGE	142.4	100%	99%	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	166.1	99%	96%	Fish-eye disease, 136120 Norum disease, 245900
LCT	145.1	99%	98%	Lactase deficiency, congenital, 223000
LDHA	69.6	94%	89%	Glycogen storage disease XI, 612933
LDHB	103.6	97%	90%	[Lactate dehydrogenase-B deficiency], 614128
LFNG	99.3	84%	82%	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LIPA	128	97%	95%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	130.1	100%	99%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LMBRD1	96.5	93%	88%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	151.2	99%	96%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	124.6	100%	99%	Majeed syndrome, 609628
LPL	175.7	100%	99%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LRAT	345.6	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	62.2	78%	65%	Leukotriene C4 synthase deficiency, 614037
LYST	171	98%	96%	Chediak-Higashi syndrome, 214500
MAN1B1	162.6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	134.6	98%	95%	Mannosidosis, alpha-, types I and II, 248500
MANBA	153.6	99%	98%	Mannosidosis, beta, 248510
MAOA	130.2	100%	99%	Brunner syndrome, 300615 {Antisocial behavior},300615
MAT1A	206.1	99%	97%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850

MCCC1	176.8	100%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	159.5	100%	99%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	134.1	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	168.5	99%	97%	Mucolipidosis IV, 252650
MFSD8	143.4	100%	99%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	177.1	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	165.6	99%	97%	Thyroid carcinoma, follicular, 188470
MLYCD	94.5	95%	91%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	222.3	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	118.2	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	227.3	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94.4	90%	80%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	92.7	98%	92%	Molybdenum cofactor deficiency A, 252150
MOCS2	183	99%	99%	Molybdenum cofactor deficiency B, 252160
MOGS	128	99%	98%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	129.4	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPI	142	100%	100%	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	57.3	93%	82%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	159.4	99%	98%	{Abruptio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MTHFR	161.7	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTM1	108.7	98%	93%	Myotubular myopathy, X-linked, 310400
MTMR2	130	100%	99%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	174.9	99%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	152.5	99%	99%	Homocystinuria-megaloblastic anemia, cbl E type, 236270

				{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	154.5	99%	98%	Methylmalonic aciduria, mut(0) type, 251000
MVK	167.2	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
NAGA	159.7	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	131.8	94%	91%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	84	97%	92%	N-acetylglutamate synthase deficiency, 237310
NEU1	20.4	72%	43%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	144.4	100%	99%	Leber congenital amaurosis 9, 608553
NNT	166.9	99%	97%	Glucocorticoid deficiency 4, 614736
NPC1	153.4	99%	99%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	148.6	100%	99%	Niemann-pick disease, type C2, 607625
NSD1	181.1	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	196.4	100%	99%	CHILD syndrome, 308050 CK syndrome, 300831
NT5C3A	80.9	92%	82%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	198.7	100%	99%	Calcification of joints and arteries, 211800
OAT	96.2	80%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	140.5	99%	98%	Dent disease 2, 300555 Lowe syndrome, 309000
OPA3	125.8	99%	97%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	124.4	99%	98%	5-oxoprolinase deficiency, 260005
OTC	128.2	99%	99%	Ornithine transcarbamylase deficiency, 311250
OXCT1	146.5	99%	99%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050

PAH	186.6	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	178.4	99%	98%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	166.1	99%	97%	Pyruvate carboxylase deficiency, 266150
PCBD1	129.9	99%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	120.1	98%	93%	Propionicacidemia, 606054
PCCB	167.8	98%	96%	Propionicacidemia, 606054
PCK1	137.1	100%	99%	?Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency, 261680
PCK2	208.3	100%	99%	PEPCK deficiency, mitochondrial, 261650
PEPD	127	99%	98%	Prolidase deficiency, 170100
PEX1	139.9	98%	97%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	124.7	97%	92%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	103.8	100%	99%	Peroxisome biogenesis disorder 14B, 614920
PEX12	157.9	100%	99%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	240	100%	99%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	145.5	99%	98%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	146.3	97%	93%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	118.8	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	187.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	87.2	100%	99%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	129.6	99%	97%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	132.1	99%	98%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	95.9	91%	85%	Heimler syndrome 2, 616617

				Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	152.7	90%	87%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	165.3	100%	99%	Glycogen storage disease VII, 232800
PGAM2	177.3	100%	100%	Glycogen storage disease X, 261670
PGAP2	173.9	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	57.8	92%	80%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	149.5	100%	99%	Congenital disorder of glycosylation, type It, 614921
PHGDH	138	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	120.6	99%	95%	Muscle glycogenosis, 300559
PHKA2	119.9	99%	99%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	147.7	100%	99%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	114.6	99%	96%	No OMIM phenotype
PHYH	90	98%	93%	Refsum disease, 266500
PIGA	97	94%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGL	125.2	100%	99%	CHIME syndrome, 280000
PIGM	159.5	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	130.2	99%	94%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	143.9	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	195.8	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	168	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	155.4	100%	99%	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501

				Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3R1	158.1	99%	98%	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	104.3	90%	87%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	117.8	100%	99%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	177.4	99%	98%	Corneal fleck dystrophy, 121850
PIP5K1C	131.7	96%	94%	Lethal congenital contractural syndrome 3, 611369
PKLR	195.6	100%	99%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PLA2G5	130.5	100%	100%	[Fleck retina, familial benign], 228980
PLA2G6	135.5	99%	97%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	158.1	100%	99%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLCB1	173.5	100%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	139.4	99%	97%	Auriculocondylar syndrome 2, 614669
PLCD1	122.7	99%	98%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	166.9	99%	98%	Nephrotic syndrome, type 3, 610725
PLCG2	130.6	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLIN1	89.6	98%	90%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	155.4	99%	96%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	143.1	97%	92%	Bruck syndrome 2, 609220
PLOD3	117.6	99%	99%	Lysyl hydroxylase 3 deficiency, 612394
PMM2	171.3	100%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	187.5	100%	99%	Pancreatic lipase deficiency, 614338
PNMT	104.5	99%	96%	?Hypertension, essential, 145500
PNP	147.3	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	139.3	99%	96%	Neutral lipid storage disease with myopathy, 610717

PNPLA6	150.4	99%	98%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
PNPO	80.9	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLR3A	153	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	163.8	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	136.7	99%	97%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMGNT2	258.7	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	223.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094
POMT1	184.3	99%	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 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	117.5	99%	96%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PPM1K	180	100%	99%	?Maple syrup urine disease, mild variant, 615135
PPOX	104.3	99%	97%	Porphyria variegata, 176200
PPT1	203.5	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	146.1	99%	95%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRODH	96.4	89%	83%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	178.9	100%	100%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PSAP	129.6	99%	99%	Combined SAP deficiency, 611721

				Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.4	91%	77%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSPH	164.6	99%	98%	Phosphoserine phosphatase deficiency, 614023
PTEN	169.6	100%	99%	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTGIS	149.3	98%	95%	Hypertension, essential, 145500
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTS	133.3	99%	96%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	103.5	99%	95%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	177.1	100%	100%	Glycogen storage disease VI, 232700
PYGM	156.7	100%	100%	McArdle disease, 232600
QDPR	91.4	100%	99%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RBCK1	112	98%	95%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	102.7	99%	94%	Leber congenital amaurosis 13, 612712
RDH5	174.3	100%	99%	Fundus albipunctatus, 136880
RFT1	114.8	99%	98%	Congenital disorder of glycosylation, type In, 612015
RPE65	165.8	100%	99%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794

RPIA	138.2	95%	92%	?Ribose 5-phosphate isomerase deficiency, 608611
SARDH	141.5	93%	91%	[Sarcosinemia], 268900
SAT1	153.9	99%	97%	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SC5D	237.6	99%	99%	Lathosterolosis, 607330
SCARB2	141.5	100%	99%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	138	100%	98%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SEPSECS	198.4	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	139.3	99%	96%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	140.9	96%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SI	125.9	98%	94%	Sucrase-isomaltase deficiency, congenital, 222900
SLC16A1	180.7	99%	98%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	142.3	99%	95%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A5	171.6	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC25A1	90.7	98%	92%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	138.2	99%	96%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	238.4	98%	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	79.7	99%	96%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	109.7	100%	99%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A32	150.2	100%	100%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	124.7	99%	97%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC2A1	191.5	100%	99%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A2	205.9	100%	99%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC30A10	194.2	100%	99%	Hypermanganesemia with dystonia 1, 613280

SLC33A1	152.6	98%	94%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	169.7	100%	99%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	244.4	99%	98%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	139.2	100%	100%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A14	120.5	99%	97%	Hypermanganesemia with dystonia 2, 617013
SLC39A4	93.7	99%	96%	Acrodermatitis enteropathica, 201100
SLC39A8	160.7	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	205.3	100%	99%	Cystinuria, 220100
SLC46A1	103.2	99%	95%	Folate malabsorption, hereditary, 229050
SLC52A1	231.4	100%	100%	Riboflavin deficiency, 615026
SLC52A2	206.8	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	108.8	99%	99%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	149.1	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	139.8	100%	99%	Renal glucosuria, 233100
SLC6A8	59	91%	81%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	114.5	100%	99%	Lysinuric protein intolerance, 222700
SLC7A9	144.1	99%	99%	Cystinuria, 220100
SLCO1B1	65.1	95%	91%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	65.3	97%	87%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	151.8	99%	98%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	79.4	89%	77%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	150.2	100%	99%	Amyotrophic lateral sclerosis 1, 105400
SPR	194	99%	90%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	131.7	99%	96%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	174.4	100%	99%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	96.1	99%	97%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	172	100%	99%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST3GAL3	195.5	100%	99%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090

ST3GAL5	144.8	95%	94%	Amish infantile epilepsy syndrome, 609056
STAR	147.6	100%	100%	Lipoid adrenal hyperplasia, 201710
STS	104	99%	95%	Ichthyosis, X-linked, 308100
SUCLA2	78.2	94%	86%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	120	100%	99%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	71	94%	83%	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SUMF1	137.1	99%	94%	Multiple sulfatase deficiency, 272200
SUOX	221.3	100%	100%	Sulfite oxidase deficiency, 272300
TALDO1	153.4	100%	99%	Transaldolase deficiency, 606003
TANGO2	158.8	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TAT	140.9	100%	100%	Tyrosinemia, type II, 276600
TAZ	123.4	99%	98%	Barth syndrome, 302060
TBXAS1	170.1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	127.8	96%	89%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	201.5	100%	100%	Transcobalamin II deficiency, 275350
TECR	108.3	99%	98%	Mental retardation, autosomal recessive 14, 614020
TH	87.6	97%	93%	Segawa syndrome, recessive, 605407
TK2	105.4	94%	90%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM165	136.2	99%	98%	Congenital disorder of glycosylation, type IIk, 614727
TMEM5	138.9	96%	93%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMLHE	110.9	99%	96%	{Autism,susceptibility to,X-linked 6}, 300872
TPI1	104.9	98%	95%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPMT	57.1	96%	86%	{Thiopurines,poor metabolism of,1}, 610460
TPP1	155.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREH	169.1	99%	94%	Trehalase deficiency, 612119
TUSC3	166.8	100%	99%	Mental retardation, autosomal recessive 7, 611093
TYMP	104.7	98%	89%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	205.9	100%	100%	Albinism, oculocutaneous, type IA, 203100

				Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYRP1	200.1	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UGT1A1	263	100%	100%	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMPS	204.3	100%	99%	Orotic aciduria, 258900
UPB1	184.6	100%	100%	Beta-ureidopropionase deficiency, 613161
UROC1	164	100%	99%	?Urocanase deficiency, 276880
UROD	174	99%	97%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	126.7	100%	100%	Porphyria, congenital erythropoietic, 263700
XDH	118.7	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.
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 14th 2017
This list is accurate for panel version DG 2.9

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