METABOLIC DISORDERS GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AASS	147.3	99%	97%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	103	100%	99%	GABA-transaminase deficiency, 613163
ABCD1	96	77%	68%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	155.2	99%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	147.9	99%	98%	Sitosterolemia, 210250
ABCG8	166.1	99%	95%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	114.9	97%	91%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	265.7	99%	99%	Chanarin-Dorfman syndrome, 275630
ACACA	155.4	98%	97%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	149.3	99%	99%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	154	99%	96%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADM	112.6	98%	94%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	139.9	99%	97%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	138.4	99%	94%	2-methylbutyrylglycinuria, 610006
ACADVL	128.3	99%	97%	VLCAD deficiency, 201475
ACAT1	138.1	98%	93%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	188.5	100%	100%	?ACAT2 deficiency, 614055
ACO2	138.6	96%	92%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	186.3	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	146.5	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	127.6	96%	91%	Mental retardation, X-linked 63, 300387
ACY1	155.9	99%	98%	Aminoacylase 1 deficiency, 609924
ADA	118	99%	97%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCY5	144.8	93%	91%	Dyskinesia, familial, with facial myokymia, 606703

ADK	106.1	98%	93%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	205.9	100%	99%	Adenylosuccinase deficiency, 103050
AGA	154.1	100%	100%	Aspartylglucosaminuria, 208400
AGK	137.6	99%	97%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	164.6	99%	97%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	120.1	98%	92%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	57.3	93%	82%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	149.5	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHCY	138.6	100%	99%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	130.7	99%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	117	98%	94%	Reticular dysgenesis, 267500
AKR1D1	113.1	97%	93%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	115	100%	99%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	107.4	99%	96%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	143	100%	99%	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	124.9	93%	89%	Microphthalmia, isolated 8, 615113
ALDH2	133.5	99%	99%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}
ALDH3A2	157.4	100%	99%	Sjogren-Larsson syndrome, 270200
ALDH4A1	130.3	99%	98%	Hyperprolinemia, type II, 239510
ALDH5A1	92.3	90%	82%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	149.3	100%	99%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	83.1	97%	89%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	191.8	99%	96%	Glycogen storage disease XII, 611881
ALDOB	174.3	99%	98%	Fructose intolerance, 229600

ALG1	60.3	53%	49%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	282.1	100%	100%	{Long QT syndrome, acquired, reduced susceptibility to}, 613688
ALG11	182.7	100%	99%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	167.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	107.4	98%	94%	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	119.8	100%	99%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	125.8	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	104.1	95%	92%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	153.6	96%	93%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	136.2	99%	98%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALOX12B	148.4	99%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	163.1	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMACR	161.6	99%	98%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	69.2	86%	75%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD3	146	99%	98%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	174	100%	99%	Glycine encephalopathy, 605899
AP1S1	121.3	100%	99%	MEDNIK syndrome, 609313
APOC2	111.6	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APRT	68.4	99%	99%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	172.1	100%	100%	Argininemia, 207800
ARSA	111.5	100%	99%	Metachromatic leukodystrophy, 250100
ARSB	144.1	98%	95%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	141	98%	92%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	118.4	99%	98%	Argininosuccinic aciduria, 207900
ASPA	151.7	99%	92%	Canavan disease, 271900
ASS1	106.2	97%	92%	Citrullinemia, 215700
ATIC	134.7	99%	98%	AICA-ribosiduria due to ATIC deficiency, 608688

ATP6V0A2	159.8	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	157.3	99%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	173	99%	99%	Wilson disease, 277900
ATP8B1	169.2	95%	93%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
AUH	89.3	98%	92%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	127.9	100%	98%	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	134.9	92%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALTL	113.5	96%	92%	Peters-plus syndrome, 261540
B3GAT3	101	99%	95%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GNT1	126	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B4GALT1	132.4	99%	99%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	116.3	96%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BAAT	140.3	98%	95%	Hypercholanemia, familial, 607748
BCKDHA	186.9	99%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	124.4	89%	81%	Maple syrup urine disease, type Ib, 248600
BCMO1	184.8	100%	100%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	145.3	99%	99%	Hyperbiliverdinemia, 614156
BMP2	204.4	99%	99%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200
BPGM	126	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	163.8	100%	99%	Biotinidase deficiency, 253260
C1GALT1C1	155.5	99%	99%	Tn polyagglutination syndrome, somatic, 300622
C7orf10	139.2	95%	90%	Glutaric aciduria III, 231690
CANT1	157.6	100%	100%	Desbuquois dysplasia 1, 251450
CAT	186	100%	99%	Acatalasemia, 614097
CBS	127.1	97%	92%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200

CEL	143.7	85%	83%	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	108.1	97%	92%	Retinitis pigmentosa 26, 608380
CERS3	139.7	100%	99%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	142	98%	96%	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	125.8	99%	96%	[Chitotriosidase deficiency], 614122
CHKB	101.2	99%	96%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	190.5	96%	94%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	93	99%	96%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	328.3	100%	100%	Macular corneal dystrophy, 217800
CHSY1	148.3	96%	94%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	125.9	98%	94%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	163.1	98%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	142.3	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	252.4	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	152.7	96%	95%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COG1	136	99%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	139	100%	99%	Congenital disorder of glycosylation, type IIj, 613489
COG5	117.8	97%	93%	Congenital disorder of glycosylation, type IIi, 613612
COG6	92.4	93%	85%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	138.9	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	124.6	99%	96%	Congenital disorder of glycosylation, type IIh, 611182
COMT	218.2	100%	99%	{Panic disorder, susceptibility to}, 167870 {Schizophrenia, susceptibility to}, 181500
CP	141	94%	90%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290

CPOX	124.8	91%	85%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	169.3	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	192.2	100%	98%	CPT deficiency, hepatic, type IA, 255120
CPT2	168.6	98%	96%	CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 Myopathy due to CPT II deficiency, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CTH	188.9	99%	99%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTNS	138.1	100%	100%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTSA	149.8	99%	99%	Galactosialidosis, 256540
CTSC	148.6	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	183.6	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	117.2	100%	99%	Pycnodysostosis, 265800
CUBN	141.9	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CYB5R3	184.9	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	141.1	99%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	182.2	99%	99%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	185.8	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	146.2	100%	99%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110

CYP19A1	206.6	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	132.1	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	15.3	56%	30%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	188.7	97%	95%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	137.1	99%	98%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	151	96%	91%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	140.9	95%	92%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	106.9	95%	90%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	147.9	97%	95%	D-2-hydroxyglutaric aciduria, 600721
DAO	144.9	100%	100%	{Schizophrenia}, 181500
DBH	153.5	100%	99%	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]
DBT	122.5	97%	92%	Maple syrup urine disease, type II, 248600
DCXR	168.4	99%	95%	[Pentosuria], 260800
DDC	118.6	99%	96%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	166.1	96%	94%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	129.1	99%	99%	?Congenital disorder of glycosylation, type Ir, 614507
DGAT1	168.9	90%	87%	?Diarrhea 7, 615863
DGKE	157.9	98%	93%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	134.6	99%	98%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	203	100%	99%	Desmosterolosis, 602398
DHCR7	176.8	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHFR	55	90%	73%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	111.8	100%	99%	Miller syndrome, 263750
DLD	142	99%	97%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	171.2	99%	97%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC19	105.3	97%	90%	3-methylglutaconic aciduria, type V, 610198
DNM1L	131.5	99%	96%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	143.2	98%	96%	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	129	99%	98%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	141	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	201.8	99%	99%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	134.5	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	136.9	89%	84%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	115.2	100%	99%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	165.1	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPYD	177.9	95%	94%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	144.7	99%	98%	Dihydropyrimidinuria, 222748
EBP	101.3	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	128	100%	99%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL4	104.8	99%	97%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
ENO3	202	100%	99%	?Glycogen storage disease XIII, 612932
EPHX1	138.4	98%	96%	Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 ?Fetal hydantoin syndrome {Preeclampsia, susceptibility to}, 189800
EPHX2	129.1	100%	99%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
ETFA	165.2	100%	99%	Glutaric acidemia IIA, 231680
ETFB	128.1	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	116.1	99%	98%	Glutaric acidemia IIC, 231680
ETHE1	86.4	99%	94%	Ethylmalonic encephalopathy, 602473
EXT1	106.1	99%	97%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	197.4	99%	99%	Exostoses, multiple, type 2, 133701

				?Seizures, scoliosis, and macrocephaly syndrome, 616682
FA2H	108.9	94%	88%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	173.1	100%	99%	Tyrosinemia, type I, 276700
FBP1	128.6	100%	99%	Fructose-1,6-bisphosphatase deficiency, 229700
FECH	142.4	99%	99%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FH	175.4	92%	88%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	93	99%	98%	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	157.4	98%	93%	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
FMO3	177.1	100%	99%	Trimethylaminuria, 602079
FOLR1	166.3	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	99.6	94%	88%	Glutamate formiminotransferase deficiency, 229100
FUCA1	156.2	99%	99%	Fucosidosis, 230000
FUT2	186.4	100%	100%	[Bombay phenotype] {Norwalk virus infection, resistance to} {Vitamin B12 plasma level QTL1}, 612542
FUT6	169.5	100%	99%	Fucosyltransferase 6 deficiency, 613852
G6PC	219.4	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	138.4	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	138	99%	98%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	126.1	100%	99%	Glycogen storage disease II, 232300
GAD1	132.5	99%	97%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	112.3	97%	93%	Krabbe disease, 245200
GALE	168.6	100%	100%	Galactose epimerase deficiency, 230350
GALK1	117.5	98%	96%	Galactokinase deficiency with cataracts, 230200

GALNS	105.4	99%	94%	Mucopolysaccharidosis IVA, 253000
GALT	168	100%	100%	Galactosemia, 230400
GAMT	119.1	97%	91%	Cerebral creatine deficiency syndrome 2, 612736
GATM	174.8	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GBA	237.3	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	187.3	100%	99%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	171.8	99%	97%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	148.6	93%	91%	Glutaricaciduria, type I, 231670
GCH1	91.2	95%	86%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	155.4	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	150.4	99%	98%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	132.5	98%	93%	{Myocardial infarction, susceptibility to}, 608446
GCSH	38.8	83%	62%	Glycine encephalopathy, 605899
GFPT1	160.1	99%	97%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GK	51.7	77%	62%	Glycerol kinase deficiency, 307030
GLA	87.1	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	93.9	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	90.1	91%	84%	Glycine encephalopathy, 605899

GLRA1	136.5	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRX5	102.2	93%	86%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	82.4	94%	86%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	101.8	99%	97%	Glutamine deficiency, congenital, 610015
GLYCTK	239	99%	99%	D-glyceric aciduria, 220120
GM2A	143.6	100%	99%	GM2-gangliosidosis, AB variant, 272750
GMPPB	256.1	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	121	97%	92%	No OMIM phenotype Leukemia, acute myelogenous, 601626
GNE	176.5	99%	99%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	180	99%	97%	Glycine N-methyltransferase deficiency, 606664
GNPAT	159.6	98%	95%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	192	98%	97%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	156.3	95%	89%	Mucolipidosis III gamma, 252605
GNS	123	96%	91%	Mucopolysaccharidosis type IIID, 252940
GOT1	140	100%	98%	Aspartate aminotransferase, serum level of, QTL1, 614419
GPD1	116.2	99%	97%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	173.4	100%	99%	Brugada syndrome 2, 611777
GPHN	192.1	98%	97%	Molybdenum cofactor deficiency C, 615501
GPI	159.4	100%	99%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPX1	38.2	87%	74%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	120.1	84%	79%	Hyperoxaluria, primary, type II, 260000
GSS	113.1	100%	99%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GUSB	125.1	90%	87%	Mucopolysaccharidosis VII, 253220
GYG1	145.7	99%	98%	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507
GYS1	122.6	99%	98%	Glycogen storage disease 0, muscle, 611556
GYS2	183.5	98%	94%	Glycogen storage disease 0, liver, 240600

H6PD	177.8	99%	99%	Cortisone reductase deficiency 1, 604931
HADH	120.8	97%	95%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	90.1	95%	90%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	100.9	94%	85%	Trifunctional protein deficiency, 609015
HAGH	139.3	99%	96%	[Glyoxalase II deficiency], 614033
HEXA	143.7	100%	99%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	152.6	97%	91%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	155	99%	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	143.1	99%	98%	Alkaptonuria, 203500
HGSNAT	116.9	82%	80%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	108.2	92%	90%	No OMIM phenotype
HIBCH	76.2	91%	72%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	160.1	99%	99%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	193.1	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	117.7	100%	99%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	152.4	99%	99%	HMG-CoA lyase deficiency, 246450
HMGCS2	149.9	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	142.1	97%	90%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HOGA1	154.2	100%	97%	Hyperoxaluria, primary, type III, 613616

HPD	151.2	100%	99%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	75.3	94%	84%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	68.9	93%	84%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD11B1	190.2	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	166.6	87%	84%	Apparent mineralocorticoid excess, 218030
HSD17B10	120.3	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B3	172	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	110.3	94%	91%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	178.2	100%	100%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	154.3	99%	93%	Bile acid synthesis defect, congenital, 1, 607765
HYAL1	125	100%	100%	?Mucopolysaccharidosis type IX, 601492
IDH2	108.8	99%	98%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	186.6	100%	100%	Retinitis pigmentosa 46, 612572
IDS	119.1	99%	98%	Mucopolysaccharidosis II, 309900
IDUA	116.9	91%	85%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IMPAD1	152.6	99%	99%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	60.2	92%	81%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
INPP5E	105.1	96%	91%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	136.5	98%	95%	Opsismodysplasia, 258480
ISPD	120	95%	85%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
IVD	126	99%	99%	Isovaleric acidemia, 243500
KMT2A	173.8	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage Wiedemann-Steiner syndrome, 605130
KMT2D	162.1	99%	99%	Kabuki syndrome 1, 147920
L2HGDH	139.8	98%	96%	L-2-hydroxyglutaric aciduria, 236792

LAMP2	134.3	92%	91%	Danon disease, 300257
LARGE	145.9	99%	98%	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	160.7	99%	96%	Fish-eye disease, 136120 Norum disease, 245900
LCT	146.7	99%	98%	Lactase deficiency, congenital, 223000
LDHA	60.1	92%	86%	Glycogen storage disease XI, 612933
LDHB	115.9	97%	86%	[Lactate dehydrogenase-B deficiency], 614128
LFNG	95.6	84%	82%	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LIPA	129.1	96%	94%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	134.6	99%	99%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LMBRD1	81.3	89%	81%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	144.7	99%	94%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	129.6	99%	99%	Majeed syndrome, 609628
LPL	183.3	100%	99%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LRAT	324	100%	99%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	60.5	79%	64%	Leukotriene C4 synthase deficiency, 614037
LYST	151.3	97%	94%	Chediak-Higashi syndrome, 214500
MAN1B1	156.2	99%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	137.2	98%	96%	Mannosidosis, alpha-, types I and II, 248500
MANBA	144.2	99%	95%	Mannosidosis, beta, 248510
MAOA	142.8	100%	99%	Brunner syndrome, 300615 {Antisocial behavior},300615
MAT1A	196.4	99%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	169.5	99%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200

MCCC2	142	99%	98%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	110.4	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	166.6	98%	96%	Mucolipidosis IV, 252650
MFSD8	137.1	99%	98%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	168.8	100%	99%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	166.8	98%	94%	Thyroid carcinoma, follicular, 188470
MLYCD	93.2	94%	91%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	201.9	99%	99%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	121.2	99%	99%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	205.3	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	82.8	87%	74%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	92.2	98%	93%	Molybdenum cofactor deficiency A, 252150
MOCS2	156.7	99%	98%	Molybdenum cofactor deficiency B, 252160
MOGS	121.3	99%	98%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	131.9	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPI	137.4	100%	100%	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	45.6	87%	75%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	157.8	99%	97%	{Abruptio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MTHFR	153.2	100%	99%	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	115.4	97%	89%	Myotubular myopathy, X-linked, 310400
MTMR2	130.1	99%	98%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	158.9	99%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	143	99%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634

MUT	130.1	99%	95%	Methylmalonic aciduria, mut(0) type, 251000
MVK	146.1	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
NAGA	162.6	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	123.7	93%	91%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	80.6	97%	91%	N-acetylglutamate synthase deficiency, 237310
NEU1	19.8	73%	44%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	143.8	99%	97%	Leber congenital amaurosis 9, 608553
NNT	157	99%	97%	Glucocorticoid deficiency 4, 614736
NPC1	162.1	99%	98%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	144	100%	99%	Niemann-pick disease, type C2, 607625
NSD1	172.3	100%	99%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	205.2	99%	99%	CHILD syndrome, 308050 CK syndrome, 300831
NT5C3A	66.5	87%	74%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	192.1	100%	99%	Calcification of joints and arteries, 211800
OAT	94.1	76%	68%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	152.2	99%	97%	Dent disease 2, 300555 Lowe syndrome, 309000
OPA3	122	99%	96%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	114.7	99%	98%	5-oxoprolinase deficiency, 260005
OTC	143.9	100%	99%	Ornithine transcarbamylase deficiency, 311250
OXCT1	129.1	99%	97%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	191.3	100%	100%	Phenylketonuria, 261600

				[Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	177.5	99%	96%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	162.8	99%	97%	Pyruvate carboxylase deficiency, 266150
PCBD1	124.7	99%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	116.6	96%	91%	Propionicacidemia, 606054
PCCB	160.5	97%	95%	Propionicacidemia, 606054
PEPD	121.4	99%	98%	Prolidase deficiency, 170100
PEX1	123.4	97%	95%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	118.3	97%	93%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	123.3	99%	99%	Peroxisome biogenesis disorder 14B, 614920
PEX12	165.4	100%	99%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	212.2	99%	98%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	143.6	99%	98%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	138.5	96%	92%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	121	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	169.7	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	86.5	99%	99%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.8	98%	94%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	126.7	99%	97%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	92	90%	84%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	138.5	89%	85%	Peroxisome biogenesis disorder 9B, 614879

				Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	175.3	100%	99%	Glycogen storage disease VII, 232800
PGAM2	168.7	100%	100%	Glycogen storage disease X, 261670
PGAP2	186.1	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	60	92%	81%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	143.1	99%	99%	Congenital disorder of glycosylation, type It, 614921
PHGDH	138.6	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	132.8	98%	96%	Muscle glycogenosis, 300559
PHKA2	130.1	100%	99%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHYH	86.5	98%	92%	Refsum disease, 266500
PIGA	102.1	92%	84%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGL	136.6	100%	98%	CHIME syndrome, 280000
PIGM	160.8	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	128.6	95%	89%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	140.6	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	174.6	99%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	171.1	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	136.5	99%	98%	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	144	99%	96%	Immunodeficiency 36, 616005 SHORT syndrome, 269880

				?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	96.5	89%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	123.1	100%	99%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	161.3	99%	97%	Corneal fleck dystrophy, 121850
PIP5K1C	122.3	96%	94%	Lethal congenital contractural syndrome 3, 611369
PKLR	182.7	100%	99%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PLA2G5	133.8	100%	100%	[Fleck retina, familial benign], 228980
PLA2G6	132.4	99%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	137.2	99%	97%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLCB1	177.3	99%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	147.9	99%	95%	Auriculocondylar syndrome 2, 614669
PLCD1	124.1	99%	97%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	171.1	99%	98%	Nephrotic syndrome, type 3, 610725
PLCG2	133.6	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLIN1	92.4	97%	89%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	149.8	100%	99%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	125.3	94%	88%	Bruck syndrome 2, 609220
PLOD3	125.3	99%	98%	Lysyl hydroxylase 3 deficiency, 612394
PMM2	178.4	99%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	197.8	100%	98%	Pancreatic lipase deficiency, 614338
PNMT	96.9	98%	96%	?Hypertension, essential, 145500
PNP	148.5	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	127.6	99%	97%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	140.9	99%	98%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
PNPO	84.2	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090

POLR3A	162.2	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	168.9	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	131.9	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	281.8	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	225.3	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	181	99%	97%	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	120.5	98%	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	174.3	99%	99%	?Maple syrup urine disease, mild variant, 615135
PPOX	108.1	99%	97%	Porphyria variegata, 176200
PPT1	190.4	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRODH	95.8	88%	82%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	201.5	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	126.7	99%	98%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	49.5	90%	74%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSPH	139.8	99%	94%	Phosphoserine phosphatase deficiency, 614023
PTEN	152.4	99%	98%	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	140.1	98%	95%	Hypertension, essential, 145500
PTPN11	101.2	96%	90%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTS	128.2	97%	89%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	105.4	99%	94%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	192.4	100%	99%	Glycogen storage disease VI, 232700
PYGM	160.1	100%	99%	McArdle disease, 232600
QDPR	88.5	100%	99%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RDH12	101.9	98%	91%	Leber congenital amaurosis 13, 612712
RDH5	160.7	100%	99%	Fundus albipunctatus, 136880
RFT1	118.8	99%	97%	Congenital disorder of glycosylation, type In, 612015
RPE65	150.7	99%	99%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	138.1	94%	91%	?Ribose 5-phosphate isomerase deficiency, 608611
SARDH	132.5	92%	91%	[Sarcosinemia], 268900
SAT1	162.6	99%	98%	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SC5D	227.6	99%	98%	Lathosterolosis, 607330
SCARB2	138.9	99%	98%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	124.8	99%	96%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SEPSECS	185	99%	99%	Pontocerebellar hypoplasia type 2D, 613811

SERAC1	125.5	98%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	146.8	97%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SI	110.4	96%	90%	Sucrase-isomaltase deficiency, congenital, 222900
SLC16A1	167.2	99%	97%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	123.8	97%	93%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A5	164.5	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC25A1	84.7	97%	90%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	125.3	98%	93%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	228.7	98%	95%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	81.4	99%	95%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	115.1	100%	99%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A38	117.7	99%	96%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC2A1	183.5	100%	100%	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	189.5	100%	99%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC30A10	192.1	99%	99%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	148.5	96%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	146	99%	97%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	230	99%	97%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	141	100%	99%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	88.4	99%	96%	Acrodermatitis enteropathica, 201100
SLC3A1	192	99%	98%	Cystinuria, 220100
SLC46A1	105.4	98%	94%	Folate malabsorption, hereditary, 229050

SLC52A1	222.1	100%	100%	Riboflavin deficiency, 615026
SLC52A2	196.4	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	124.5	100%	99%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	160.8	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	136.3	100%	99%	Renal glucosuria, 233100
SLC6A8	61.1	92%	82%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	127.2	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	152.6	100%	98%	Cystinuria, 220100
SLCO1B1	53.4	92%	85%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	56.8	93%	78%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	134.3	99%	97%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	70.1	88%	76%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	179.7	100%	99%	Amyotrophic lateral sclerosis 1, 105400
SPR	183.9	97%	89%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	132.3	98%	95%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	178	100%	99%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	91.8	100%	97%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	166.9	99%	99%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST3GAL3	193.7	100%	100%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	138.4	95%	94%	Amish infantile epilepsy syndrome, 609056
STAR	134.5	99%	99%	Lipoid adrenal hyperplasia, 201710
STS	111.8	99%	96%	Ichthyosis, X-linked, 308100
SUCLA2	69.4	92%	82%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	111.3	99%	97%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	65.9	91%	79%	No OMIM phenotype
SUMF1	138.4	97%	92%	Multiple sulfatase deficiency, 272200
SUOX	219.5	100%	100%	Sulfite oxidase deficiency, 272300
TALDO1	143.2	100%	99%	Transaldolase deficiency, 606003

TANGO2	161	100%	99%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TAT	141.9	100%	100%	Tyrosinemia, type II, 276600
TAZ	126.3	100%	98%	Barth syndrome, 302060
TBXAS1	161.1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	125.6	95%	88%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	192.4	100%	100%	Transcobalamin II deficiency, 275350
TECR	96.4	99%	97%	Mental retardation, autosomal recessive 14, 614020
TH	83.6	97%	92%	Segawa syndrome, recessive, 605407
TK2	109.9	92%	87%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM165	122.2	98%	96%	Congenital disorder of glycosylation, type IIk, 614727
TMEM5	140	93%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMLHE	116.6	99%	96%	{Autism,susceptibility to,X-linked 6}, 300872
TPI1	109.8	98%	96%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPMT	49.3	92%	80%	{Thiopurines,poor metabolism of,1}, 610460
TPP1	158.7	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREH	153.5	99%	95%	Trehalase deficiency, 612119
TUSC3	153.5	99%	98%	Mental retardation, autosomal recessive 7, 611093
TYMP	96.7	96%	88%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	205.6	100%	99%	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	209.7	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UGT1A1	244.7	100%	99%	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	196.5	100%	99%	Orotic aciduria, 258900
UPB1	177.2	100%	100%	Beta-ureidopropionase deficiency, 613161
UROC1	150	100%	99%	?Urocanase deficiency, 276880
UROD	178.2	100%	99%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	119.9	100%	100%	Porphyria, congenital erythropoietic, 263700
XDH	119.9	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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