METABOLIC DISORDERS GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
AASS	101,1	100%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	66,2	100%	92%	GABA-transaminase deficiency, 613163
ABCD1	31,8	73%	64%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	100,4	100%	97%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	135,1	99%	87%	Sitosterolemia, 210250
ABCG8	90,7	97%	96%	Sitosterolemia, 210250 Gallbladder disease 4, 611465
ABHD12	61	98%	83%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	121,2	100%	100%	Chanarin-Dorfman syndrome, 275630
ACACA	97,1	99%	98%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	92,1	99%	94%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	91	100%	99%	ACAD9 deficiency, 611126
ACADM	147,3	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	107,7	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	85,7	98%	96%	2-methylbutyrylglycinuria, 610006
ACADVL	87,5	100%	98%	VLCAD deficiency, 201475

ACAT1	109,9	100%	98%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	93,4	100%	100%	ACAT2 deficiency, 614055
ACO2	77,9	90%	83%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	75,3	99%	93%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	77,6	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	67,4	100%	97%	Mental retardation, X-linked 63, 300387
ACY1	87,8	100%	97%	Aminoacylase 1 deficiency, 609924
ADA	70,7	100%	95%	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700
ADCY5	83,2	98%	94%	Dyskinesia, familial, with facial myokymia, 606703
ADK	123,9	94%	94%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	127,7	100%	98%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AGA	117,2	100%	89%	Aspartylglucosaminuria, 208400
AGK	106,6	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	145,6	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	62,9	95%	82%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	116,3	100%	100%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	88,5	97%	90%	Hyperoxaluria, primary, type 1, 259900
AHCY	70,6	92%	73%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	86,6	100%	98%	Hemolytic anemia due to adenylate kinase deficiency, 612631

AK2	73,5	77%	75%	Reticular dysgenesis, 267500
AKR1D1	98	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	83,5	100%	95%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	41	87%	79%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	91,9	97%	91%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	83,5	99%	93%	Microphthalmia, isolated 8, 615113
ALDH2	87,4	98%	93%	Alcohol sensitivity, acute, 610251 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to} {Esophageal cancer, alcohol-related, susceptibility to}
ALDH3A2	97,3	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	69,4	94%	89%	Hyperprolinemia, type II, 239510
ALDH5A1	71,8	97%	95%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100,9	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	64,9	90%	87%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	111,4	98%	94%	Glycogen storage disease XII, 611881
ALDOB	106,5	100%	98%	Fructose intolerance, 229600
ALG1	47,7	45%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	135,9	100%	98%	{Acquired long QT syndrome, reduced susceptibility to}, 613688
ALG11	154,3	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	103,4	100%	97%	Congenital disorder of glycosylation, type Ig, 607143

ALG13	58,2	93%	85%	Congenital disorder of glycosylation, type Is, 300884
ALG2	115	100%	97%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	83,7	100%	93%	Congenital disorder of glycosylation, type Id, 601110
ALG6	104,1	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	85,2	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	83,5	100%	99%	Congenital disorder of glycosylation, type Il, 608776
ALOX12B	101,5	100%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	80,7	100%	100%	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300 Hypophosphatasia, adult, 146300
AMACR	82,8	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	65,8	89%	85%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD3	85,5	99%	97%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	127,5	100%	99%	Glycine encephalopathy, 605899
AP1S1	79,9	100%	99%	MEDNIK syndrome, 609313
APOC2	156,8	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APRT	47,5	100%	91%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	135	98%	91%	Argininemia, 207800
ARSA	87,1	97%	94%	Metachromatic leukodystrophy, 250100
ARSB	91,1	100%	99%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200

ASAH1	99,1	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	81,3	99%	96%	Argininosuccinic aciduria, 207900
ASPA	112,8	100%	99%	Canavan disease, 271900
ASS1	40,4	85%	60%	Citrullinemia, 215700
ATIC	115,4	100%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP6V0A2	106,5	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	60,3	100%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	125,7	100%	98%	Wilson disease, 277900
ATP8B1	113,5	99%	98%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480
AUH	114,5	100%	100%	3-methylglutaconic aciduria, type I, 250950
B3GALTL	107,6	95%	95%	Peters-plus syndrome, 261540
B3GAT3	58,8	97%	83%	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600
B3GNT1	95,8	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B3GNT2	128,4	100%	100%	No OMIM phenotype
B4GALT1	78,9	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	89,7	100%	95%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
BAAT	123	100%	99%	Hypercholanemia, familial, 607748

BCKDHA	102,4	100%	98%	Maple syrup urine disease, type Ia, 248600
BCKDHB	95,9	98%	84%	Maple syrup urine disease, type Ib, 248600
BCMO1	131,9	100%	99%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	85,8	100%	99%	Hyperbiliverdinemia, 614156
BMP2	115,8	100%	100%	{HFE hemochromatosis, modifier of}, 235200 Brachydactyly, type A2, 112600
BPGM	148,3	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	141,7	100%	100%	Biotinidase deficiency, 253260
C1GALT1C1	86,6	100%	99%	Tn polyagglutination syndrome, somatic, 300622
CANT1	93,1	99%	97%	Desbuquois dysplasia, 251450
CAT	91,7	98%	91%	Acatalasemia, 614097
CBS	73,6	98%	89%	subtelomeric Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CEL	57,3	64%	61%	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	137,2	100%	100%	Retinitis pigmentosa 26, 608380
CERS3	84,7	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023

CFTR	123,1	95%	95%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF {Pancreatitis, idiopathic}, 167800 {Hypertrypsinemia, neonatal} {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400
CHKB	91,1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	130,2	100%	100%	Ehlers-Danlos syndrome, musculocontractural type , 601776
CHST3	67,2	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	123,3	100%	100%	Macular corneal dystrophy, 217800
CHSY1	147,3	98%	97%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	86,5	100%	99%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139,7	100%	90%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	63,7	98%	81%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	133,8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
COG1	121	100%	98%	Congenital disorder of glycosylation, type IIg, 611209
COG4	86,4	98%	95%	Congenital disorder of glycosylation, type IIj, 613489
COG5	101,6	97%	94%	Congenital disorder of glycosylation, type IIi, 613612
COG6	101,9	97%	95%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	77	100%	96%	Congenital disorder of glycosylation, type IIe, 608779
COG8	116,5	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COMT	77,9	100%	100%	{Schizophrenia, susceptibility to}, 181500 {Panic disorder, susceptibility to}, 167870

CP	90,4	99%	92%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPOX	85,2	100%	97%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	107,5	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	96,1	100%	98%	CPT deficiency, hepatic, type IA, 255120
CPT2	109,4	92%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CTH	130,2	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated -3
CTNS	114,9	97%	94%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800 -3
CTSA	92,8	100%	98%	Galactosialidosis, 256540
CTSC	86,4	100%	100%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CTSD	96,2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	129,9	100%	100%	Pycnodysostosis, 265800
CUBN	88,4	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
CYB5R3	77,9	97%	94%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	89,8	100%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743

CYP11B1	128	98%	94%	anti-Lepore-like Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	104,9	98%	91%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 {Low renin hypertension, susceptibility to} Aldosterone to renin ratio raised
CYP17A1	105,5	99%	96%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	131	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300 -3
CYP1B1	103,3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	5,2	17%	10%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	118,1	100%	96%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	107,1	100%	98%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	107,1	99%	96%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	108,4	100%	99%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	97,1	100%	96%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	57,7	96%	86%	D-2-hydroxyglutaric aciduria, 600721
DAO	106,8	100%	100%	{Schizophrenia}, 181500 (2)
DBH	99	100%	97%	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360
DBT	111,7	100%	100%	Maple syrup urine disease, type II, 248600
DCXR	93,1	100%	100%	Pentosuria, 260800
DDC	88,8	100%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643

DDHD1	141,9	100%	97%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	97,6	100%	98%	Congenital disorder of glycosylation, type Ir, 614507
DGAT1	87,8	91%	88%	?Diarrhea 7, 615863
DGKE	111,9	100%	99%	Nephrotic syndrome, type 7, 615008
DGUOK	99,3	100%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	86,6	99%	97%	Desmosterolosis, 602398
DHCR7	115,9	100%	97%	Smith-Lemli-Opitz syndrome, 270400
DHFR	51,9	81%	58%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	100,6	100%	99%	Miller syndrome, 263750
DLD	143,7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	116,6	97%	96%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC19	57,8	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNM1L	103	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	80	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Lethal congenital contracture syndrome 5, 615368

DNMT1	101,5	99%	96%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	93,8	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DPAGT1	90,9	99%	95%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	157,7	100%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	59,9	98%	88%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	101,1	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPYD	119,1	98%	96%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	60,6	100%	98%	Dihydropyrimidinuria, 222748
EBP	43,9	94%	73%	Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	64	98%	92%	No OMIM phenotype
ELOVL4	102,9	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	107,7	99%	96%	Glycogen storage disease XIII, 612932
EPHX1	94	95%	91%	?Fetal hydantoin syndrome Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 {Preeclampsia, susceptibility to}, 189800
EPHX2	94,4	95%	94%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
ETFA	115,4	100%	100%	Glutaric acidemia IIA, 231680
ETFB	107,8	100%	99%	Glutaric acidemia IIB, 231680

ETFDH	132,6	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	61	100%	96%	Ethylmalonic encephalopathy, 602473
EXT1	104,7	98%	95%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	117,3	100%	96%	Exostoses, multiple, type 2, 133701
FA2H	64,6	87%	76%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	108,6	100%	98%	Tyrosinemia, type I, 276700
FBP1	90,3	100%	96%	Fructose-1,6-bidphosphatase deficiency, 229700
FECH	106,9	100%	98%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FH	85,7	98%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	80,2	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	108,3	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FMO3	107,4	100%	100%	Trimethylaminuria, 602079
FOLR1	78,4	100%	97%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	58,1	92%	81%	Glutamate formiminotransferase deficiency, 229100

FUCA1	74,3	100%	96%	Fucosidosis, 230000
FUT2	211,3	100%	100%	{Norwalk virus infection, resistance to} [Bombay phenotype] {Vitamin B12 plasma level QTL1}, 612542
FUT6	77	81%	73%	Fucosyltransferase 6 deficiency, 613852
G6PC	140,5	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	116,3	100%	100%	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541
G6PD	57	95%	92%	Hemolytic anemia due to G6PD deficiency Favism, 134700 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	96,6	100%	98%	Glycogen storage disease II, 232300
GAD1	96,2	100%	98%	Cerebral palsy, spastic quadriplegic, 1, 603513
GAD1	96,2	100%	98%	Immunoglobulin A deficiency (2)
GALC	96,4	100%	96%	Krabbe disease, 245200
GALE	115,6	100%	100%	Galactose epimerase deficiency, 230350
GALK1	96,4	100%	100%	Galactokinase deficiency with cataracts, 230200
GALNS	67,4	92%	92%	Mucopolysaccharidosis IVA, 253000
GALT	113,2	100%	100%	Galactosemia, 230400
GAMT	91,8	98%	92%	Cerebral creatine deficiency syndrome 2, 612736
GATM	88,8	100%	94%	Cerebral creatine deficiency syndrome 3, 612718

GBA	62,1	63%	58%	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013 {Parkinson disease, late-onset, susceptibility to}, 168600 {Lewy body dementia, susceptibility to}, 127750
GBE1	104,3	98%	94%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	80,6	92%	91%	Glutaricaciduria, type I, 231670
GCH1	99,7	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	82,5	100%	97%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, gestational, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176
GCLC	137,3	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	92,8	84%	84%	{Myocardial infarction, susceptibility to}, 608446
GCSH	14,8	52%	39%	Glycine encephalopathy, 605899
GFPT1	105,6	100%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GK	24,5	77%	56%	Glycerol kinase deficiency, 307030
GLA	47,2	95%	86%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	76,5	99%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010

GLDC	60,1	98%	85%	Glycine encephalopathy, 605899
GLRA1	113,9	100%	98%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRX5	29,9	72%	46%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	111,8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	28,1	60%	49%	Glutamine deficiency, congenital, 610015
GLYCTK	83,8	99%	98%	D-glyceric aciduria, 220120
GM2A	127,8	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPB	118,5	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351
GMPS	116,3	100%	100%	Leukemia, acute myelogenous, 601626
GNE	99,5	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GNMT	85,8	100%	99%	Glycine N-methyltransferase deficiency, 606664
GNPAT	115,9	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	135,6	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNS	80	94%	85%	Mucopolysaccharidosis type IIID, 252940
GOT1	100,1	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GPD1	72,9	100%	99%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	105,6	100%	100%	Brugada syndrome 2, 611777
GPHN	119,4	100%	100%	Molybdenum cofactor deficiency, type C, 252150

GPI	91,9	100%	97%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPX1	19,7	75%	55%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	88,2	97%	78%	Hyperoxaluria, primary, type II, 260000
GSS	80	98%	94%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GUSB	61,9	89%	81%	Mucopolysaccharidosis VII, 253220
GYG1	45,5	83%	60%	Glycogen storage disease XV, 613507
GYS1	66,9	95%	82%	Glycogen storage disease 0, muscle, 611556
GYS2	92,1	100%	100%	Glycogen storage disease, type 0, 240600
H6PD	125,6	100%	100%	Cortisone reductase deficiency 1, 604931
HADH	91,5	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	93	96%	87%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	94,9	100%	99%	Trifunctional protein deficiency, 609015
HAGH	82,9	99%	97%	[Glyoxalase II deficiency], 614033
HEXA	90,7	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	114,8	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800

HFE	101,2	100%	98%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferrin serum level QTL2], 614193
HGD	89,4	100%	100%	Alkaptonuria, 203500
HGSNAT	105,4	93%	93%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	130,2	99%	92%	No OMIM phenotype
HIBCH	67,3	100%	99%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	116,1	100%	100%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	141,8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	99,6	99%	98%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	103,5	100%	99%	HMG-CoA lyase deficiency, 246450
HMGCS2	114,9	100%	99%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	65,9	100%	95%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HOGA1	72,6	100%	94%	Hyperoxaluria, primary, type III, 613616
HPD	98,3	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	50,8	100%	78%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HS6ST1	7,3	14%	0%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD11B1	109,1	100%	100%	Cortisone reductase deficiency 2, 614662

HSD11B2	113,2	78%	78%	Apparent mineralocorticoid excess, 218030
HSD17B10	54,1	95%	91%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B3	105	99%	96%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	98,2	100%	99%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	44	93%	78%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	72,7	92%	82%	Bile acid synthesis defect, congenital, 1, 607765
HYAL1	97,4	99%	96%	Mucopolysaccharidosis type IX, 601492
IDH2	106,8	100%	94%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	116,8	95%	95%	Retinitis pigmentosa 46, 612572
IDS	55,1	84%	77%	telomeric IDS2 source of inversion in IDS Mucopolysaccharidosis II, 309900
IDUA	86	95%	84%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IMPAD1	130,4	100%	100%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	35,8	83%	60%	pseudogene on 16p13.13 Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
INPP5E	74,2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPPL1	94,9	98%	95%	Opsismodysplasia, 258480
ISPD	79,5	93%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
IVD	96	100%	96%	Isovaleric acidemia, 243500
L2HGDH	81,2	100%	97%	L-2-hydroxyglutaric aciduria, 236792

LAMP2	61,4	98%	90%	Danon disease, 300257
LARGE	108,7	99%	96%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LCAT	107,1	93%	88%	very close to HP Norum disease, 245900 Fish-eye disease, 136120
LCT	130	100%	99%	Lactase deficiency, congenital, 223000
LDHA	45,9	76%	58%	Glycogen storage disease XI, 612933
LDHB	89,5	100%	100%	Lactate dehydrogenase-B deficiency, 614128
LFNG	58,5	84%	79%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LIPA	109,4	100%	100%	GOT Wolman disease, 278000 Cholesteryl ester storage disease, 278000 -3
LIPC	84	100%	95%	[High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025
LMBRD1	108,9	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	100,3	100%	99%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	75,5	100%	97%	Majeed syndrome, 609628
LPL	109,4	100%	100%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]
LRAT	196,5	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	64	87%	66%	Leukotriene C4 synthase deficiency, 614037
LYST	120,5	99%	96%	Chediak-Higashi syndrome, 214500
MAN1B1	96,6	100%	99%	Mental retardation, autosomal recessive 15, 614202

MAN2B1	80,2	97%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	92,3	100%	99%	Mannosidosis, beta, 248510
MAOA	53,7	100%	95%	Brunner syndrome, 300615
MAT1A	94,5	100%	99%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	97	99%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	113,1	98%	92%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	79,6	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	96,7	97%	92%	Mucolipidosis IV, 252650
MFSD8	120	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	217,2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	165,5	100%	100%	Thyroid carcinoma, follicular, 188470
MLYCD	78,3	94%	81%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	174,5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	79,8	100%	92%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	177,9	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400

MMADHC	79,5	100%	100%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MOCS1	68,9	98%	92%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	116,2	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MPDU1	113,2	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPI	92,1	100%	97%	Congenital disorder of glycosylation, type Ib, 602579
MTHFR	93,8	100%	98%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTM1	55,5	100%	98%	Myotubular myopathy, X-linked, 310400
MTMR2	106,9	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	104,3	100%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	105,2	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	124,8	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MVK	88,3	100%	97%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
NAGA	78,6	100%	99%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	67,1	94%	89%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	52,2	85%	75%	N-acetylglutamate synthase deficiency, 237310

NEU1	13,5	64%	20%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	96	100%	100%	Leber congenital amaurosis 9, 608553
NNT	96,8	100%	100%	Glucocorticoid deficiency 4, 614736
NPC1	90,2	99%	98%	{Nasopharyngeal carcinoma 1} (2)
NPC2	74,4	100%	100%	Niemann-pick disease, type C2, 607625
NSD1	124,8	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650
NSDHL	52,1	97%	94%	CHILD syndrome, 308050 CK syndrome, 300831
NT5E	111,7	100%	100%	Calcification of joints and arteries, 211800
OAT	44,9	83%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	59,4	97%	94%	Lowe syndrome, 309000 Dent disease 2, 300555
OPA3	108	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	97,3	99%	96%	5-oxoprolinase deficiency, 260005
OTC	54,5	100%	94%	Ornithine transcarbamylase deficiency, 311250
OXCT1	96,3	100%	98%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	86,5	100%	98%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	112,1	90%	86%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PC	96,7	95%	92%	Pyruvate carboxylase deficiency, 266150
PCCA	96,2	96%	94%	Propionicacidemia, 606054

PCCB	114,2	100%	100%	pccB complementation group Propionicacidemia, 606054
PEPD	66,4	97%	89%	closely linked to APOC2 Prolidase deficiency, 170100
PEX1	124,5	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73	95%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	157,2	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	133	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	134,6	100%	95%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	92,3	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX14	92,3	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	91,8	94%	84%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	104,7	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	159,1	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	113	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	141,1	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	88,7	97%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	91,3	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863

PEX7	102,2	99%	93%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	102,2	100%	100%	Glycogen storage disease VII, 232800
PGAM2	96,8	100%	100%	Glycogen storage disease X, 261670
PGAP2	123,4	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	42,7	75%	67%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	109,2	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHGDH	84,6	100%	97%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	47,3	94%	88%	Muscle glycogenosis, 300559
PHKA2	49,6	97%	85%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHYH	88,8	100%	99%	Refsum disease, 266500
PIGA	66,6	99%	98%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGL	95,7	100%	100%	CHIME syndrome, 280000
PIGM	105,4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	107	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	104,6	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	140,2	100%	99%	Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	182,7	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300

PIK3CA	126,7	93%	90%	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Nonsmall cell lung cancer, somatic, 211980 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, somatic, 603387 Cowden syndrome 5, 615108
PIK3R1	144,1	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	80,8	96%	87%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	72,9	100%	98%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	138	100%	100%	Corneal fleck dystrophy, 121850
PIP5K1C	58,1	87%	83%	Lethal congenital contractural syndrome 3, 611369
PKLR	123,2	100%	97%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PLA2G5	108,2	100%	100%	Fleck retina, familial benign, 228980
PLA2G6	77,6	99%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLA2G7	122,6	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLCB1	113	100%	98%	Epileptic encephalopathy, early infantile, 12, 613722

PLCB4	93,3	100%	98%	Auriculocondylar syndrome 2, 614669
PLCD1	95,1	97%	92%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	123,8	99%	97%	Nephrotic syndrome, type 3, 610725
PLCG2	107,3	99%	98%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLIN1	54,5	90%	78%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	78,4	100%	96%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	111	100%	100%	Bruck syndrome 2, 609220
PLOD3	79,6	95%	85%	Lysyl hydroxylase 3 deficiency, 612394
PMM2	92,4	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	86,7	100%	100%	Pancreatic lipase deficiency, 614338
PNMT	46,4	95%	92%	?Hypertension, essential, 145500
PNP	124,9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	78,5	100%	91%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	81,5	100%	96%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	71,8	100%	97%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLR3A	90,6	99%	96%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	104,2	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381

POMGNT1	96,7	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMT1	99,9	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	74,5	100%	93%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PPM1K	99,8	100%	94%	Maple syrup urine disease, mild variant, 615135
PPOX	90,4	100%	97%	Porphyria variegata, 176200
PPT1	66,2	100%	93%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRODH	51,5	84%	66%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	63,6	99%	97%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PSAP	80,2	100%	98%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSAT1	41,1	67%	54%	Phosphoserine aminotransferase deficiency, 610992
PSPH	46,7	78%	44%	Phosphoserine phosphatase deficiency, 614023

PTEN	137,7	100%	97%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Thyroid carcinoma, follicular, somatic, 188470 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355
PTGIS	56,1	100%	89%	Hypertension, essential, 145500
PTPN11	46,9	88%	66%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTS	118,4	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	83,6	100%	96%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	102,8	100%	99%	Glycogen storage disease VI, 232700
PYGM	97,3	100%	98%	McArdle disease, 232600
QDPR	77,2	100%	98%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RDH12	63,6	91%	80%	Leber congenital amaurosis 13, 612712
RDH5	107,3	100%	100%	Fundus albipunctatus, 136880
RFT1	78,3	100%	97%	Congenital disorder of glycosylation, type In, 612015
RPE65	114,7	100%	98%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794

RPIA	72	100%	95%	Ribose 5-phosphate isomerase deficiency, 608611
SARDH	71,8	97%	92%	[Sarcosinemia], 268900
SAT1	74,9	100%	96%	Keratosis follicularis spinulosa decalvans, 308800
SC5D	175,4	100%	100%	Lathosterolosis, 607330
SCARB2	88,9	100%	95%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	98,4	99%	97%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SEPSECS	104,5	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	89,8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	76	94%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SI	107,2	100%	99%	Sucrase-isomaltase deficiency, congenital, 222900
SLC16A1	139,9	100%	99%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021
SLC17A5	100,9	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A5	123,3	100%	99%	Carnitine deficiency, systemic primary, 212140
SLC25A1	73	89%	82%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	99	100%	99%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	100,6	88%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC25A19	71,1	100%	97%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	75,1	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A38	74,4	100%	96%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950

SLC2A1	94,3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC2A2	125,4	100%	100%	{Diabetes mellitus, noninsulin-dependent} Fanconi-Bickel syndrome, 227810
SLC30A10	133,4	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	107,9	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	117,6	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	97,8	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	86,8	99%	97%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	65,8	100%	98%	Acrodermatitis enteropathica, 201100
SLC3A1	134	100%	100%	Cystinuria, 220100
SLC46A1	79,2	100%	98%	Folate malabsorption, hereditary, 229050
SLC52A1	132	100%	100%	Riboflavin deficiency, 615026
SLC52A2	116,3	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	69,1	100%	95%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	106,2	100%	99%	Glucose/galactose malabsorption, 606824
SLC5A2	81,4	98%	95%	Renal glucosuria, 233100
SLC6A8	4,4	13%	5%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	90,3	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	69,4	100%	96%	Cystinuria, 220100

SLCO1B1	113,3	100%	99%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	121,6	100%	97%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	109,7	99%	91%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	12,7	54%	29%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	100,5	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SPR	67,2	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	79,2	95%	90%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	110	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	66,8	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	130,2	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST3GAL3	114	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
STAR	100,6	100%	100%	Lipoid adrenal hyperplasia, 201710
STS	69,1	98%	92%	nonlyonizing Ichthyosis, X-linked, 308100
SUCLA2	81,6	94%	91%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	94,7	95%	91%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	76,2	93%	92%	No OMIM phenotype
SUMF1	69,1	100%	95%	Multiple sulfatase deficiency, 272200
SUOX	172	100%	100%	Sulfite oxidase deficiency, 272300
TALDO1	91,9	100%	100%	Transaldolase deficiency, 606003

TAT	100,6	100%	100%	Tyrosinemia, type II, 276600
TAZ	50,6	100%	98%	Barth syndrome, 302060
TBXAS1	99,9	100%	99%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	78,3	95%	85%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	103,4	100%	97%	Transcobalamin II deficiency, 275350
TECR	81,1	100%	89%	Mental retardation, autosomal recessive 14, 614020
TH	77,8	96%	83%	Segawa syndrome, recessive, 605407
TK2	89,7	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM165	92,6	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM5	168,4	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMLHE	37,7	86%	77%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TPI1	68	100%	97%	Hemolytic anemia due to triosephosphate isomerase deficiency
TPMT	95,7	100%	100%	6-mercaptopurine sensitivity, 610460
TPP1	135,3	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREH	104,8	94%	89%	Trehalase deficiency, 612119
TUSC3	126,6	100%	99%	Mental retardation, autosomal recessive 7, 611093
TYMP	91,2	99%	92%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	132,9	74%	74%	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800

				[Skin/hair/eye pigmentation 3, blue/green eyes], 601800
TYRP1	126	100%	100%	Albinism, oculocutaneous, type III, 203290 Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair), 612271
UGT1A1	146,5	100%	98%	Crigler-Najjar syndrome, type I, 218800 [Gilbert syndrome], 143500 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transcient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816
UMPS	111,2	100%	100%	Orotic aciduria, 258900
UPB1	110,4	100%	100%	Beta-ureidopropionase deficiency, 613161
UROC1	73,3	98%	83%	Urocanase deficiency, 276880
UROD	84,5	97%	90%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	71,7	99%	89%	Porphyria, congenital erythropoietic, 263700
XDH	89,2	100%	99%	Xanthinuria, type I, 278300

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors