METABOLIC DISORDERS GENE PANEL DG 2.16 (643 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AASS	131,5	99.9%	99.4%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	83,2	99.9%	98.3%	GABA-transaminase deficiency, 613163
ABCC8	125,8	100.0%	99.9%	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD1	87,4	77.2%	75.0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	164,3	100.0%	99.7%	No OMIM phenotype
ABCD3	108,1	99.5%	97.3%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	129	99.8%	98.4%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	139,5	100.0%	99.9%	Sitosterolemia, 210250
ABCG8	133,9	99.7%	98.2%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	93,1	100.0%	98.9%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	180,9	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACACA	109,9	98.3%	97.5%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	122,1	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	124,3	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	129,5	99.8%	99.2%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	148,2	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	113,1	99.9%	97.3%	2-methylbutyrylglycinuria, 610006
ACADVL	115,8	99.8%	98.0%	VLCAD deficiency, 201475
ACAT1	110,2	99.7%	98.3%	Alpha-methylacetoacetic aciduria, 203750

ACAT2	132,1	100.0%	100.0%	?ACAT2 deficiency, 614055
ACBD5	145,1	99.6%	98.0%	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACO2	115,3	95.8%	89.5%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	123,7	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	111	100.0%	99.3%	Bile acid synthesis defect, congenital, 6, 617308
ACSF3	145,8	99.9%	99.1%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	102,8	98.6%	93.5%	Mental retardation, X-linked 63, 300387
ACY1	118,5	100.0%	98.6%	Aminoacylase 1 deficiency, 609924
ADA	104,6	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	128,6	100.0%	99.7%	No OMIM phenotype
ADCY5	131,8	97.8%	94.7%	Dyskinesia, familial, with facial myokymia, 606703
ADK	102,4	99.8%	98.0%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	138,6	99.2%	98.6%	Adenylosuccinase deficiency, 103050
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGK	108,5	99.5%	95.7%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146,9	100.0%	99.4%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	162,6	99.1%	94.8%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	75,4	99.5%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	160,8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHCY	111,6	99.9%	97.7%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	136,9	100.0%	99.9%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	96,1	98.7%	94.4%	Reticular dysgenesis, 267500
AKR1D1	91,5	99.6%	95.7%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	94,9	99.5%	94.7%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740

ALAS2	74,7	98.9%	94.7%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	113,7	100.0%	99.8%	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	102,4	99.7%	97.0%	Microphthalmia, isolated 8, 615113
ALDH2	126,5	100.0%	100.0%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}, 0
ALDH3A2	113,5	95.3%	94.3%	Sjogren-Larsson syndrome, 270200
ALDH4A1	123,9	100.0%	99.8%	Hyperprolinemia, type II, 239510
ALDH5A1	91	99.3%	93.2%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	104,9	100.0%	99.6%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	66,7	93.5%	86.1%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	120,4	76.5%	74.5%	Glycogen storage disease XII, 611881
ALDOB	135,3	100.0%	99.3%	Fructose intolerance, hereditary, 229600
ALG1	46,5	53.2%	50.2%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	274,1	100.0%	100.0%	{Long QT syndrome, acquired, reduced susceptibility to}, 613688
ALG11	129,3	96.8%	96.3%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	155,7	100.0%	99.9%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	77,3	98.5%	92.1%	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG14	199,4	100.0%	100.0%	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	103,2	100.0%	100.0%	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ALG6	101,6	99.1%	95.6%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118,5	96.6%	96.2%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874

ALG9	113	100.0%	99.6%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALOX12B	125,6	100.0%	99.8%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	154,8	100.0%	99.7%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMACR	157,7	100.0%	100.0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	101,5	98.1%	90.6%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	115,8	99.9%	98.6%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	117,2	99.9%	98.9%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	142,7	100.0%	100.0%	Glycine encephalopathy, 605899
AP1S1	101	100.0%	99.8%	MEDNIK syndrome, 609313
APOC2	103,7	100.0%	100.0%	Hyperlipoproteinemia, type Ib, 207750
APRT	93,2	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	159,1	100.0%	100.0%	Argininemia, 207800
ARSA	138,5	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSB	109,4	99.9%	98.9%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	125,7	99.3%	97.2%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	123,6	100.0%	98.5%	Argininosuccinic aciduria, 207900
ASPA	116,1	99.7%	96.9%	Canavan disease, 271900
ASS1	97,4	95.0%	87.1%	Citrullinemia, 215700
ATIC	113,9	100.0%	99.7%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A1	111,1	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP6AP1	105,6	99.8%	96.9%	Immunodeficiency 47, 300972
ATP6V0A2	117,4	99.9%	99.0%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V1A	133,4	99.6%	97.3%	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1E1	66,5	92.3%	85.9%	Cutis laxa, autosomal recessive, type IIC, 617402

ATP7A	111,2	99.5%	96.7%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	128,7	99.9%	99.1%	Wilson disease, 277900
ATP8B1	114	97.5%	94.6%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
AUH	127	100.0%	99.7%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	124,4	100.0%	99.6%	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	93,9	92.9%	91.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	81,7	82.6%	77.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	121	99.6%	96.5%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	96,6	99.7%	99.1%	Peters-plus syndrome, 261540
B4GALT1	112,2	99.8%	97.7%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	123,9	99.8%	98.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	136,9	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	103	99.8%	97.3%	Hypercholanemia, familial, 607748
BCKDHA	176,9	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	123,3	98.6%	92.8%	Maple syrup urine disease, type Ib, 248600
BCO1	129,2	100.0%	100.0%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	111,9	100.0%	99.9%	Hyperbiliverdinemia, 614156
BMP2	163,4	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BPGM	101,3	100.0%	100.0%	Erythrocytosis, familial, 8, 222800
BTD	126,6	99.9%	99.7%	Biotinidase deficiency, 253260
C1GALT1C1	139,9	100.0%	99.0%	Tn polyagglutination syndrome, somatic, 300622

CA5A	93,2	99.6%	95.7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	136,7	99.9%	99.2%	Epileptic encephalopathy, early infantile, 50, 616457
CANT1	144,9	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAT	136,9	100.0%	100.0%	Acatalasemia, 614097
CBS	123,3	99.9%	99.0%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCDC115	77,9	88.9%	87.1%	Congenital disorder of glycosylation, type IIo, 616828
CEL	146,5	94.0%	90.4%	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	114,1	99.4%	97.2%	Retinitis pigmentosa 26, 608380
CERS3	95,2	99.8%	98.2%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	113,5	99.4%	97.4%	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800
CHIT1	110,6	99.7%	98.0%	[Chitotriosidase deficiency], 614122
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	160,6	99.9%	98.9%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	133,8	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	279,8	100.0%	100.0%	Macular corneal dystrophy, 217800
CHSY1	125,9	99.3%	97.9%	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	114,7	92.6%	91.9%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	138,7	99.9%	98.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	132,3	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	144,5	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COG1	108,4	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209

COG2	124,6	99.7%	98.1%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	94,5	100.0%	99.6%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	126,3	99.9%	98.4%	Congenital disorder of glycosylation, type IIi, 613612
COG6	90,4	99.1%	96.0%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	106,1	100.0%	99.7%	Congenital disorder of glycosylation, type IIe, 608779
COG8	145	100.0%	98.5%	Congenital disorder of glycosylation, type IIh, 611182
COMT	161,8	100.0%	99.9%	{Panic disorder, susceptibility to}, 167870 {Schizophrenia, susceptibility to}, 181500
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	168,1	100.0%	100.0%	No OMIM phenotype Cerebellar ataxia and static encephalopmyopathy (Malicdan (2018) Hum Mutat 39,69) Intellectual disability (Najmabadi (2011) Nature 478,57)
COQ6	127,5	99.9%	98.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	138,3	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	99,5	100.0%	99.8%	Nephrotic syndrome, type 9, 615573
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	100,6	93.1%	87.4%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPOX	134,1	99.5%	97.2%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	133,8	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	131,5	99.9%	98.4%	CPT deficiency, hepatic, type IA, 255120

CPT2	139,2	98.3%	98.2%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CTH	141	100.0%	99.8%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTNS	112,6	100.0%	99.5%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTSA	132,9	100.0%	99.9%	Galactosialidosis, 256540
CTSC	116,2	100.0%	100.0%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	171	99.8%	97.8%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	86,3	100.0%	99.8%	Pycnodysostosis, 265800
CUBN	103,2	99.6%	97.6%	Megaloblastic anemia-1, Finnish type, 261100
CYB561	145	92.8%	92.7%	Orthostatic hypotension 2, 618182
CYB5R3	152,1	99.2%	98.3%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	121,2	99.2%	95.0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	155,9	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	156	100.0%	100.0%	Aldosterone to renin ratio raised, 0 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}, 0
CYP17A1	108,5	100.0%	99.6%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	125,7	99.4%	97.3%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300

CYP1B1	134,4	100.0%	100.0%	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP21A2	91,6	99.2%	93.4%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	173	100.0%	99.7%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	147,1	100.0%	99.7%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	130,7	99.7%	96.8%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	134,3	98.4%	95.5%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	103,8	99.6%	96.6%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	142	100.0%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAO	105,1	100.0%	99.7%	{Schizophrenia}, 181500
DBH	145,6	100.0%	99.9%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	109,9	99.6%	96.9%	Maple syrup urine disease, type II, 248600
DCXR	169,1	100.0%	99.9%	[Pentosuria], 260800
DDC	97,9	99.5%	95.0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	161,6	99.9%	98.4%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	114	100.0%	99.8%	?Congenital disorder of glycosylation, type Ir, 614507
DGAT1	150,3	96.7%	92.0%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	127,8	99.8%	98.3%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	119,4	99.9%	97.9%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	155,8	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861

DHFR	50	94.1%	83.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	98,8	100.0%	100.0%	Miller syndrome, 263750
DLD	119,2	99.9%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	134,7	100.0%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	140,7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	93,8	98.4%	92.3%	3-methylglutaconic aciduria, type V, 610198
DNM1L	119,5	99.9%	98.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	123,9	99.7%	96.7%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	114,3	99.2%	98.7%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	116,4	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	157,2	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	87,5	100.0%	99.9%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	134,7	95.2%	88.2%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	88,5	99.8%	97.6%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	200,5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPYD	141,6	99.5%	96.4%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	117,5	100.0%	99.8%	Dihydropyrimidinuria, 222748
DTYMK	108,5	100.0%	100.0%	No OMIM phenotype
EBP	63,2	99.5%	95.2%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	103,8	100.0%	99.7%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL1	85,9	99.8%	97.5%	No OMIM phenotype

ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ENO3	173,2	100.0%	100.0%	?Glycogen storage disease XIII, 612932
EOGT	102,7	79.3%	78.1%	Adams-Oliver syndrome 4, 615297
EPHX1	116	99.2%	96.0%	?Hypercholanemia, familial, 607748
EPHX2	97,7	99.6%	97.4%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
ETFA	132,7	100.0%	99.8%	Glutaric acidemia IIA, 231680
ETFB	116,8	100.0%	100.0%	Glutaric acidemia IIB, 231680
ETFDH	114,4	100.0%	99.3%	Glutaric acidemia IIC, 231680
ETHE1	97,3	99.9%	97.8%	Ethylmalonic encephalopathy, 602473
EXT1	88,6	99.6%	98.0%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	118	99.9%	99.1%	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	128,4	100.0%	99.8%	Tyrosinemia, type I, 276700
FAR1	73,7	97.2%	91.8%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBP1	105,5	100.0%	99.3%	Fructose-1,6-bisphosphatase deficiency, 229700
FDFT1	136,3	100.0%	99.6%	Squalene synthase deficiency, 618156
FECH	104	100.0%	99.7%	Protoporphyria, erythropoietic, 1, 177000
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	153,3	100.0%	100.0%	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	107,5	99.7%	96.1%	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	170,7	100.0%	99.6%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	131,9	100.0%	99.3%	Trimethylaminuria, 602079
FOLR1	107,4	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	116,8	98.7%	95.2%	Glutamate formiminotransferase deficiency, 229100
FUCA1	125,9	100.0%	99.9%	Fucosidosis, 230000
FUK	NC	NC	NC	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FUT2	154,5	100.0%	100.0%	[Bombay phenotype, digenic], 616754 {Norwalk virus infection, resistance to}, 0 {Vitamin B12 plasma level QTL1}, 612542
FUT6	142,5	100.0%	100.0%	Fucosyltransferase 6 deficiency, 613852
G6PC	146,8	100.0%	99.9%	Glycogen storage disease Ia, 232200
G6PC3	114,6	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	114,4	99.5%	97.4%	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	160,8	100.0%	99.9%	Glycogen storage disease II, 232300
GAD1	112,7	99.9%	99.7%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	102,9	99.8%	98.8%	Krabbe disease, 245200
GALE	140	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALK1	165,2	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALM	91,9	100.0%	99.9%	No OMIM phenotype
GALNS	108,3	100.0%	99.3%	Mucopolysaccharidosis IVA, 253000
GALNT3	125,8	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	152,6	100.0%	100.0%	Galactosemia, 230400
GAMT	112,5	98.3%	91.5%	Cerebral creatine deficiency syndrome 2, 612736

GANAB	107,1	99.9%	98.3%	Polycystic kidney disease 3, 600666
GATM	137,3	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GBA	169,8	100.0%	100.0%	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	141,5	100.0%	99.6%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	157,4	99.9%	99.7%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	145,9	100.0%	99.1%	Glutaricaciduria, type I, 231670
GCH1	84,8	100.0%	99.5%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	138,6	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	143,7	99.3%	95.8%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	101,7	99.7%	97.2%	{Myocardial infarction, susceptibility to}, 608446
GCSH	32,1	88.4%	69.8%	?Glycine encephalopathy, 605899
GFPT1	146	99.9%	99.1%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GIF	NC	NC	NC	Intrinsic factor deficiency, 261000
GK	44,1	84.9%	63.9%	Glycerol kinase deficiency, 307030
GLA	73,6	99.5%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	59,2	90.6%	79.2%	Glycine encephalopathy, 605899

GLRA1	96,8	100.0%	99.7%	Hyperekplexia 1, 149400
GLRX5	137,6	99.6%	96.1%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	65,5	98.1%	87.5%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	77	99.7%	96.5%	Glutamine deficiency, congenital, 610015
GLYCTK	161,3	100.0%	99.5%	D-glyceric aciduria, 220120
GM2A	122	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMPPA	147,2	100.0%	99.8%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	211,8	100.0%	100.0%	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	120,6	98.2%	94.2%	No OMIM phenotype Leukemia, acute myelogenous, 601626
GNE	113,8	100.0%	99.3%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	126,9	99.9%	98.7%	Glycine N-methyltransferase deficiency, 606664
GNPAT	127,2	99.5%	96.8%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	148	100.0%	99.3%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	177,6	100.0%	98.5%	Mucolipidosis III gamma, 252605
GNS	94,5	99.6%	95.2%	Mucopolysaccharidosis type IIID, 252940
GOT1	107,7	100.0%	99.4%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	80,3	95.8%	89.5%	No OMIM phenotype
GPD1	87,7	100.0%	99.4%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	128	100.0%	99.9%	Brugada syndrome 2, 611777
GPHN	144,7	99.9%	98.8%	Molybdenum cofactor deficiency C, 615501
GPI	141,4	100.0%	99.8%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPT2	121,7	100.0%	99.4%	Mental retardation, autosomal recessive 49, 616281
GPX1	50,6	99.6%	95.9%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164

GRHPR	99,6	84.2%	81.7%	Hyperoxaluria, primary, type II, 260000
GSS	93,3	100.0%	99.2%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GUSB	99,5	92.5%	90.5%	Mucopolysaccharidosis VII, 253220
GYG1	125,6	100.0%	99.4%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	109,6	100.0%	98.6%	Glycogen storage disease 0, muscle, 611556
GYS2	120,1	99.7%	98.0%	Glycogen storage disease 0, liver, 240600
H6PD	193,5	99.0%	99.0%	Cortisone reductase deficiency 1, 604931
HADH	111,1	99.3%	98.8%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	72,9	96.3%	89.3%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	77,6	96.7%	83.8%	Trifunctional protein deficiency, 609015
HAGH	139,7	100.0%	99.8%	[Glyoxalase II deficiency], 614033
HEXA	106,3	93.7%	92.4%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	163	99.7%	98.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	108	100.0%	98.9%	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	98,2	100.0%	99.8%	Alkaptonuria, 203500
HGSNAT	98,3	87.2%	86.2%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	106,3	97.3%	92.8%	No OMIM phenotype
HIBCH	69,9	96.3%	79.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	116,5	100.0%	99.7%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460

HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMBS	97,3	100.0%	98.4%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	119,4	99.9%	98.7%	HMG-CoA lyase deficiency, 246450
HMGCS2	102,4	100.0%	99.4%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	137,4	96.5%	90.7%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HOGA1	149,6	100.0%	99.1%	Hyperoxaluria, primary, type III, 613616
HPD	148,1	100.0%	99.7%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	59,8	98.3%	88.2%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	72,8	97.7%	92.0%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD11B1	113,8	100.0%	99.0%	Cortisone reductase deficiency 2, 614662
HSD11B2	165,6	94.3%	87.3%	Apparent mineralocorticoid excess, 218030
HSD17B10	92,4	100.0%	98.4%	HSD10 mitochondrial disease, 300438
HSD17B3	116,4	100.0%	99.9%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	109,4	96.3%	93.6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	131,8	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	143,7	99.5%	96.2%	Bile acid synthesis defect, congenital, 1, 607765
HTRA2	132,6	100.0%	99.6%	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
HYAL1	110,7	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492
IDH2	98,5	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	128,2	95.5%	95.4%	Retinitis pigmentosa 46, 612572
IDS	100,9	99.9%	97.1%	Mucopolysaccharidosis II, 309900
IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IMPAD1	170,4	100.0%	99.9%	Chondrodysplasia with joint dislocations, GPAPP type, 614078

IMPDH1	53,6	95.3%	84.1%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	127,9	99.8%	98.0%	Opsismodysplasia, 258480
IREB2	130,2	100.0%	99.5%	No OMIM phenotype
ISPD	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITPA	130,2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
IVD	100	100.0%	99.9%	Isovaleric acidemia, 243500
KMT2A	133	100.0%	99.9%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
L2HGDH	124	99.0%	96.7%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	92,3	97.9%	92.8%	Danon disease, 300257
LARGE1	115,2	100.0%	99.7%	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	140,7	99.4%	95.1%	Fish-eye disease, 136120 Norum disease, 245900
LCT	118,8	99.8%	97.9%	Lactase deficiency, congenital, 223000
LDHA	55,6	96.6%	88.0%	Glycogen storage disease XI, 612933
LDHB	86	94.8%	82.9%	[Lactate dehydrogenase-B deficiency], 614128
LFNG	117,6	92.8%	87.7%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	125,3	99.9%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	105	96.5%	94.4%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	98,5	100.0%	99.1%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPT1	203,2	100.0%	99.9%	Lipoyltransferase 1 deficiency, 616299

LIPT2	91,2	99.9%	99.3%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	100,1	98.9%	94.1%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	123,4	99.1%	96.4%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	97,8	100.0%	99.6%	Majeed syndrome, 609628
LPL	128,2	100.0%	99.9%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LRAT	240,7	100.0%	100.0%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	83,3	94.2%	79.5%	Leukotriene C4 synthase deficiency, 614037
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
MAN1B1	125,5	100.0%	99.8%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	128,6	99.9%	98.6%	Mannosidosis, alpha-, types I and II, 248500
MANBA	118,3	99.5%	97.5%	Mannosidosis, beta, 248510
MAOA	98,9	100.0%	99.0%	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAT1A	144,1	99.8%	98.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	137,6	100.0%	99.4%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	119	100.0%	99.7%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	121,1	100.0%	99.9%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	157,1	99.9%	99.0%	Mucolipidosis IV, 252650
MFSD2A	114,3	100.0%	99.3%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	121,3	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	145,4	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	163,8	100.0%	99.5%	{Thyroid carcinoma, follicular}, 188470
MLYCD	95,7	99.4%	96.5%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168,5	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100

MMAB	94,6	100.0%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	196	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	81,2	92.7%	79.5%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	91,2	98.8%	95.7%	Molybdenum cofactor deficiency A, 252150
MOCS2	137,7	99.6%	99.5%	Molybdenum cofactor deficiency B, 252160
MOGS	141	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	102,4	100.0%	99.6%	Congenital disorder of glycosylation, type If, 609180
MPI	110,1	100.0%	99.9%	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	51,6	95.8%	88.5%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	115,4	99.8%	97.4%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTM1	79,1	98.7%	91.9%	Myotubular myopathy, X-linked, 310400
MTMR2	99,1	99.9%	98.5%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	131,4	99.9%	99.4%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	131,1	100.0%	99.0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	NC	NC	NC	Methylmalonic aciduria, mut(0) type, 251000
MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900

NADK2	163,3	99.9%	99.0%	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	121,7	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	117,7	97.1%	94.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	104,3	100.0%	99.9%	N-acetylglutamate synthase deficiency, 237310
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NMNAT1	113,5	100.0%	98.5%	Leber congenital amaurosis 9, 608553
NNT	124,6	100.0%	98.5%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NPC1	117,8	100.0%	99.2%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	124,7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPL	111,5	100.0%	99.9%	No OMIM phenotype
NSD1	147	100.0%	99.8%	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	125,8	99.7%	97.1%	CHILD syndrome, 308050 CK syndrome, 300831
NT5C3A	64,1	97.2%	85.7%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	151,4	100.0%	99.9%	Calcification of joints and arteries, 211800
NUS1	53,3	71.5%	44.1%	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
OAT	68,2	81.7%	70.1%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	106,2	99.8%	98.3%	Dent disease 2, 300555 Lowe syndrome, 309000
OPA3	156,6	100.0%	99.2%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	141,5	100.0%	99.9%	5-oxoprolinase deficiency, 260005

OTC	111,4	100.0%	99.7%	Ornithine transcarbamylase deficiency, 311250
OXCT1	125,5	99.7%	98.2%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	126,4	100.0%	100.0%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	155,4	99.9%	98.7%	Pyruvate carboxylase deficiency, 266150
PCBD1	103,9	100.0%	99.7%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,2	99.3%	95.5%	Propionicacidemia, 606054
PCCB	111,8	99.3%	96.9%	Propionicacidemia, 606054
PCK1	119,4	100.0%	99.9%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	164,9	100.0%	100.0%	PEPCK deficiency, mitochondrial, 261650
PCYT1A	95,6	97.9%	94.4%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDSS1	104,8	96.7%	87.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	117,4	100.0%	99.6%	Prolidase deficiency, 170100
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	113,3	99.9%	97.4%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	87,9	100.0%	99.4%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	120,6	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	179,6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	130,5	99.8%	97.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	140,8	98.6%	94.8%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	84,9	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	134,9	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867

PEX26	94,3	100.0%	99.6%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	113,9	99.9%	99.2%	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	106,5	98.5%	92.0%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	113,7	100.0%	99.2%	Glycogen storage disease VII, 232800
PGAM2	163,6	100.0%	100.0%	Glycogen storage disease X, 261670
PGAP1	110,9	99.1%	95.8%	Mental retardation, autosomal recessive 42, 615802
PGAP2	134,7	100.0%	99.5%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	70,3	63.5%	59.9%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	44,7	90.9%	75.9%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	128,8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PGM3	149,3	99.9%	99.6%	Immunodeficiency 23, 615816
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	90,2	97.4%	91.6%	Muscle glycogenosis, 300559
PHKA2	93,7	99.9%	98.7%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	125,3	99.9%	99.1%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	112,6	99.9%	97.5%	No OMIM phenotype
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	85,9	99.3%	92.2%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGL	122,1	99.7%	99.6%	CHIME syndrome, 280000
PIGM	148,9	100.0%	100.0%	Glycosylphosphatidylinositol deficiency, 610293

PIGN	106,3	93.6%	91.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	144,5	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	89,3	94.8%	86.0%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGQ	134,6	94.0%	92.2%	No OMIM phenotype Intractable seizure, developmental delay, and optic atrophy (Alazami (2015) Cell Rep 10, 148) Ohtahara syndrome (Martin (2014) Hum Mol Genet 23, 3200)
PIGT	159,3	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	124,4	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	145	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	90,3	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	127,7	100.0%	99.8%	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3R1	124,3	99.9%	98.9%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R2	104,3	93.9%	90.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	120,7	100.0%	99.9%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	136,4	99.9%	99.7%	Corneal fleck dystrophy, 121850
PIP5K1C	136,6	99.8%	97.6%	Lethal congenital contractural syndrome 3, 611369
PKLR	169,2	100.0%	99.7%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200

PLA2G5	104,8	100.0%	100.0%	[Fleck retina, familial benign], 228980
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	120,9	99.8%	98.6%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLCB1	134,9	100.0%	99.7%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	102,5	99.8%	98.0%	Auriculocondylar syndrome 2, 614669
PLCD1	116,1	100.0%	99.3%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	125	99.8%	99.0%	Nephrotic syndrome, type 3, 610725
PLCG2	105,8	100.0%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLIN1	93,1	100.0%	99.3%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	131,9	99.8%	97.3%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	121,3	99.6%	97.3%	Bruck syndrome 2, 609220
PLOD3	109,7	100.0%	99.9%	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	95,3	99.6%	95.3%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	133,9	100.0%	97.7%	?Pancreatic lipase deficiency, 614338
PNMT	106	100.0%	99.8%	?Hypertension, essential, 145500
PNP	108,6	100.0%	99.5%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	142,7	100.0%	99.8%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	137,9	99.9%	99.5%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPO	74,4	100.0%	99.3%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	134,6	99.9%	99.4%	Dowling-Degos disease 2, 615327
POGLUT1	101,2	100.0%	98.7%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696

POLR3A	116,8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	129,8	99.7%	98.2%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	115,5	100.0%	99.6%	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 Retinitis pigmentosa 76, 617123
POMGNT2	201,7	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	138,7	100.0%	100.0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	130,6	99.7%	97.8%	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	103,3	100.0%	98.4%	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
PPCS	148,5	100.0%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PPM1K	132,5	100.0%	100.0%	?Maple syrup urine disease, mild variant, 615135
PPOX	95,2	99.8%	97.5%	Porphyria variegata, 176200
PPT1	136,6	90.2%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	129,9	99.7%	97.4%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKCSH	152,6	99.5%	94.8%	Polycystic liver disease 1, 174050

PRODH	81,8	89.0%	81.7%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	111,6	100.0%	99.9%	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	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	42,8	90.3%	72.5%	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSPH	126,6	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PTEN	129,7	99.6%	97.0%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174
PTGIS	113,7	99.5%	96.4%	Hypertension, essential, 145500
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTS	101,5	99.8%	98.4%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	96	99.7%	97.4%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	141	100.0%	100.0%	Glycogen storage disease VI, 232700
PYGM	121,1	100.0%	99.9%	McArdle disease, 232600
QDPR	97,9	100.0%	99.2%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RBCK1	107,9	100.0%	99.2%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	80,7	99.8%	97.2%	Leber congenital amaurosis 13, 612712
RDH5	167,7	100.0%	100.0%	Fundus albipunctatus, 136880
RFT1	105,7	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015

RPE65	131,9	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	113	100.0%	98.8%	?Ribose 5-phosphate isomerase deficiency, 608611
SARDH	129,1	92.9%	91.4%	[Sarcosinemia], 268900
SAT1	122,5	100.0%	98.9%	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SC5D	153,6	99.8%	99.3%	Lathosterolosis, 607330
SCARB2	105,8	99.8%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	107,8	99.7%	96.4%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	146,2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	131	99.8%	99.0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SELENBP1	121,6	100.0%	99.8%	Extraoral halitosis due to MTO deficiency, 618148
SEPSECS	159,6	100.0%	99.6%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	140,2	97.6%	94.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SI	118,7	99.4%	95.9%	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	111	99.9%	99.2%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC12A1	144,2	100.0%	99.8%	Bartter syndrome, type 1, 601678
SLC13A3	89,5	100.0%	99.5%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC16A1	138,1	100.0%	99.2%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	137,7	99.8%	96.1%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A12	117,7	100.0%	99.8%	Hypouricemia, renal, 220150
SLC22A5	129,7	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140

SLC25A1	103,2	99.3%	95.1%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	120,1	99.8%	98.9%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	146,8	97.9%	93.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	77,4	99.9%	97.8%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	90,7	100.0%	100.0%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	122,3	99.9%	99.4%	No OMIM phenotype ?Synpolydactyly (Meyertholen (2012) Mol Syndromol 3 25)
SLC25A32	128,2	100.0%	99.9%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	94,5	99.1%	95.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	130	99.9%	98.5%	No OMIM phenotype Mitochondrial myopathy (Shamseldin (2016) Hum Genet 135,21)
SLC28A1	130,4	100.0%	99.7%	No OMIM phenotype
SLC2A1	148,9	92.8%	92.8%	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	158,1	100.0%	99.9%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	104,8	100.0%	98.7%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	176,1	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	132	99.7%	97.7%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	127,1	100.0%	99.4%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	104,8	99.8%	98.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	66,6	80.6%	78.3%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	187,8	100.0%	99.8%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	125	99.5%	97.2%	Schneckenbecken dysplasia, 269250

SLC37A4	114,3	100.0%	99.6%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A14	95,4	99.9%	97.9%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	114,2	100.0%	99.0%	Acrodermatitis enteropathica, 201100
SLC39A8	140,9	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	144,5	100.0%	99.4%	Cystinuria, 220100
SLC46A1	111,1	99.9%	98.4%	Folate malabsorption, hereditary, 229050
SLC52A1	198,3	100.0%	100.0%	Riboflavin deficiency, 615026
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	118,8	100.0%	99.8%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	110,7	100.0%	99.3%	Glucose/galactose malabsorption, 606824
SLC5A2	135	100.0%	100.0%	Renal glucosuria, 233100
SLC6A19	129,3	100.0%	100.0%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A8	53,5	96.1%	83.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	105,5	100.0%	99.6%	Lysinuric protein intolerance, 222700
SLC7A9	119,8	100.0%	98.8%	Cystinuria, 220100
SLCO1B1	53,5	97.4%	89.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	50,9	97.5%	87.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	146,4	100.0%	99.2%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	63	88.9%	73.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	123,6	100.0%	100.0%	Amyotrophic lateral sclerosis 1, 105400
SPR	145,7	100.0%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	108,6	98.5%	93.4%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	142,8	100.0%	99.9%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	85,6	100.0%	98.1%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	139,9	99.8%	98.3%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SSR4	109,5	100.0%	99.7%	Congenital disorder of glycosylation, type Iy, 300934

ST3GAL3	134,7	100.0%	99.5%	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	101,8	89.0%	84.9%	Salt and pepper developmental regression syndrome, 609056
STAR	135	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
STS	78,7	99.3%	95.2%	Ichthyosis, X-linked, 308100
STT3A	123,1	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127,4	99.9%	99.6%	?Congenital disorder of glycosylation, type Ix, 615597
SUCLA2	58,8	91.7%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	102,9	99.9%	99.6%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	58,1	91.8%	79.3%	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SUGCT	127,1	99.4%	95.8%	Glutaric aciduria III, 231690
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
SUOX	167,2	100.0%	100.0%	Sulfite oxidase deficiency, 272300
TALDO1	148,2	100.0%	99.6%	Transaldolase deficiency, 606003
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	115	100.0%	100.0%	Tyrosinemia, type II, 276600
TAZ	114,5	99.3%	95.8%	Barth syndrome, 302060
TBXAS1	128,8	100.0%	100.0%	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	131,4	99.2%	96.6%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	148,5	100.0%	100.0%	Transcobalamin II deficiency, 275350
TECR	124,9	100.0%	99.6%	Mental retardation, autosomal recessive 14, 614020
TH	96,3	100.0%	98.2%	Segawa syndrome, recessive, 605407
TIMM50	122,9	99.9%	98.7%	3-methylglutaconic aciduria, type IX, 617698
TK2	103,8	100.0%	99.2%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560

TKT	115,6	98.7%	98.1%	Short stature, developmental delay, and congenital heart defects, 617044
TMEM165	148,2	100.0%	99.8%	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	118,4	100.0%	99.8%	Congenital disorder of glycosylation, type IIp, 616829
TMEM5	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM70	117,9	99.8%	97.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	87,4	99.1%	94.8%	{Autism, susceptibility to, X-linked 6}, 300872
TPI1	112,1	99.9%	96.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	94	100.0%	98.7%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPMT	42,2	96.7%	83.3%	{Thiopurines, poor metabolism of, 1}, 610460
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAK1	149,3	100.0%	99.6%	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	125,6	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC2L	198,9	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TREH	141,3	98.2%	93.2%	Trehalase deficiency, 612119
TUSC3	155,1	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TYMP	120,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	147,9	100.0%	99.9%	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	152,3	100.0%	100.0%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271

UGT1A1	184,1	100.0%	100.0%	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	149,3	100.0%	98.8%	Orotic aciduria, 258900
UPB1	143	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161
UROC1	132,8	100.0%	99.9%	?Urocanase deficiency, 276880
UROD	130,8	98.9%	95.6%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	103,8	100.0%	99.7%	Porphyria, congenital erythropoietic, 263700
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
XDH	93,8	100.0%	99.7%	Xanthinuria, type I, 278300
XYLT1	128,1	99.9%	98.2%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	147,5	99.7%	98.1%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800

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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors