METABOLIC DISORDERS GENE PANEL DG 2.17 (675 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AASS	131.3	100.0%	99.6%	Hyperlysinemia, 238700
ABAT	86.1	99.9%	98.4%	GABA-transaminase deficiency, 613163
ABCC8	134.7	100.0%	99.9%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD1	95.7	77.7%	74.9%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD2	164.9	100.0%	99.9%	No OMIM Disease ID
ABCD3	106.9	99.6%	96.7%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	139.9	99.9%	98.5%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	147.1	100.0%	99.9%	Sitosterolemia 2, 618666
ABCG8	146.0	99.9%	98.9%	Sitosterolemia 1, 210250
ABHD12	96.9	100.0%	99.5%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	183.6	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACACA	111.9	98.3%	97.6%	No OMIM disease ID
ACAD8	131.8	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	130.9	100.0%	98.8%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	124.9	99.9%	99.0%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	164.6	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	112.2	100.0%	99.1%	2-methylbutyrylglycinuria, 610006
ACADVL	125.2	99.9%	98.7%	VLCAD deficiency, 201475
ACAT1	110.0	99.9%	97.1%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	136.4	100.0%	100.0%	No OMIM disease ID
ACBD5	144.7	99.8%	98.1%	No OMIM Disease ID
ACO2	125.5	95.6%	90.3%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	129.5	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	117.5	100.0%	99.6%	Bile acid synthesis defect, congenital, 6, 617308

ACSF3	158.9	99.9%	99.4%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100.3	98.2%	92.7%	Mental retardation, X-linked 63, 300387
ACY1	128.5	99.9%	99.1%	Aminoacylase 1 deficiency, 609924
ADA	111.3	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	147.4	100.0%	99.8%	No OMIM Disease ID
ADCY5	144.1	98.7%	96.4%	Dyskinesia, familial, with facial myokymia, 606703
ADK	101.3	99.9%	97.3%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	147.2	99.2%	98.9%	Adenylosuccinase deficiency, 103050
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGK	109.6	99.6%	95.5%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	141.9	100.0%	99.7%	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	180.5	99.7%	97.0%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	74.8	100.0%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	176.8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHCY	120.8	100.0%	98.5%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	149.9	100.0%	100.0%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	100.4	98.7%	94.5%	Reticular dysgenesis, 267500
AKR1D1	92.8	99.2%	96.1%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	101.8	99.4%	95.4%	Porphyria, acute hepatic, 612740
ALAS2	77.2	99.1%	95.5%	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ALDH18A1	116.9	100.0%	99.8%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH1A3	110.3	100.0%	98.4%	Microphthalmia, isolated 8, 615113
ALDH2	136.6	100.0%	100.0%	Alcohol sensitivity, acute, 610251
ALDH3A2	116.9	95.3%	94.2%	Sjogren-Larsson syndrome, 270200
ALDH4A1	136.8	100.0%	99.8%	Hyperprolinemia, type II, 239510
ALDH5A1	95.5	99.6%	95.6%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	106.5	100.0%	99.4%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	68.0	94.2%	86.7%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	130.1	76.6%	75.2%	Glycogen storage disease XII, 611881
ALDOB	140.0	100.0%	99.1%	Fructose intolerance, hereditary, 229600

ALG1	51.3	53.6%	52.1%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	267.3	100.0%	100.0%	No OMIM Disease ID
ALG11	132.1	96.8%	96.5%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	169.5	100.0%	100.0%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	77.7	98.6%	92.4%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG14	204.6	100.0%	100.0%	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	112.6	100.0%	100.0%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	117.9	100.0%	100.0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98.1	98.9%	94.9%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118.5	96.8%	95.7%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	114.8	100.0%	99.8%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALOX12B	137.8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	168.4	99.9%	99.5%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
AMACR	168.4	100.0%	100.0%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMN	118.2	99.1%	93.0%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	115.8	99.9%	98.7%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	124.2	99.9%	99.1%	No OMIM disease ID
AMT	151.3	100.0%	100.0%	Glycine encephalopathy, 605899
AP1S1	105.8	100.0%	99.9%	MEDNIK syndrome, 609313
AP3B2	135.0	99.8%	97.9%	Epileptic encephalopathy, early infantile, 48, 617276
APOC2	112.2	100.0%	100.0%	Hyperlipoproteinemia, type Ib, 207750
APRT	105.5	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	158.2	100.0%	100.0%	Argininemia, 207800
ARSA	154.9	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSB	111.3	100.0%	99.4%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	124.7	99.6%	96.8%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	135.7	99.9%	99.2%	Argininosuccinic aciduria, 207900
ASNS	81.9	97.9%	91.0%	Asparagine synthetase deficiency, 615574

ASPA	118.0	99.9%	96.9%	Canavan disease, 271900
ASS1	106.1	95.4%	88.7%	Citrullinemia, 215700
ATIC	114.9	100.0%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A1	114.1	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP6AP1	113.6	99.8%	97.7%	Immunodeficiency 47, 300972
ATP6V0A2	120.5	100.0%	99.6%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	133.1	99.8%	97.3%	Epileptic encephalopathy, infantile or early childhood, 3, 618012 Cutis laxa, autosomal recessive, type IID, 617403
ATP6V1E1	67.0	91.8%	86.3%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	137.1	99.9%	99.3%	Wilson disease, 277900
ATP8B1	115.1	98.0%	95.0%	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
AUH	135.7	100.0%	99.8%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	126.4	100.0%	100.0%	No OMIM disease ID
B3GALNT2	94.8	93.1%	91.1%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	96.5	87.5%	80.2%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	134.2	99.9%	97.0%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	97.3	100.0%	99.7%	Peters-plus syndrome, 261540
B4GALT1	121.2	99.7%	97.4%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	138.9	100.0%	99.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	153.5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	110.3	99.7%	97.9%	Hypercholanemia, familial, 607748
BCAT1	143.5	100.0%	100.0%	No OMIM disease ID
BCAT2	159.1	100.0%	100.0%	No OMIM disease ID
BCKDHA	193.6	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	122.4	97.8%	90.2%	Maple syrup urine disease, type Ib, 248600
BCKDK	223.0	100.0%	100.0%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCO1	135.0	100.0%	100.0%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	117.6	100.0%	99.9%	Hyperbiliverdinemia, 614156

BMP2	180.6	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877
BPGM	105.8	100.0%	100.0%	Erythrocytosis, familial, 8, 222800
BTD	135.6	100.0%	99.8%	Biotinidase deficiency, 253260
C1GALT1C1	132.5	99.9%	99.0%	Tn polyagglutination syndrome, somatic, 300622
CA5A	99.0	99.9%	97.2%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	147.4	100.0%	99.6%	Epileptic encephalopathy, early infantile, 50, 616457
CANT1	158.4	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAT	141.9	100.0%	100.0%	Acatalasemia, 614097
CBS	136.4	100.0%	99.3%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCDC115	89.9	88.5%	87.0%	Congenital disorder of glycosylation, type IIo, 616828
CEL	158.1	96.1%	91.6%	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	115.2	99.5%	96.8%	Retinitis pigmentosa 26, 608380
CERS3	93.3	100.0%	98.5%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	112.9	99.4%	97.3%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHIT1	118.7	99.9%	98.5%	No OMIM disease ID
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	180.8	100.0%	99.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	146.9	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	322.9	100.0%	100.0%	Macular corneal dystrophy, 217800
CHSY1	134.8	99.8%	99.0%	Temtamy preaxial brachydactyly syndrome, 605282
CLCN7	162.0	99.9%	98.9%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLN3	123.4	92.5%	92.2%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.4	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	141.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	156.2	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	135.3	99.7%	97.4%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CMAS	93.7	99.9%	98.4%	No OMIM Disease ID
COG1	117.9	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209

COG2	126.1	99.5%	97.2%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	99.0	100.0%	99.7%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	123.1	99.8%	97.9%	Congenital disorder of glycosylation, type IIi, 613612
COG6	87.3	98.6%	95.6%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	111.8	100.0%	99.9%	Congenital disorder of glycosylation, type IIe, 608779
COG8	160.1	100.0%	98.6%	Congenital disorder of glycosylation, type IIh, 611182
COMT	177.2	100.0%	100.0%	No OMIM disease ID
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	116.2	91.7%	90.8%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	176.0	100.0%	100.0%	No OMIM Disease ID
COQ6	136.6	99.6%	97.4%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	142.8	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	177.7	100.0%	100.0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	109.0	100.0%	99.9%	Nephrotic syndrome, type 9, 615573
COQ9	78.6	99.9%	98.5%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	99.2	92.9%	87.3%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CPOX	141.1	99.7%	97.8%	Harderoporphyria, 121300 Coproporphyria, 121300
CPS1	133.4	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	135.4	99.9%	98.6%	CPT deficiency, hepatic, type IA, 255120
CPT2	152.7	98.3%	98.3%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CRAT	127.2	100.0%	100.0%	?Neurodegeneration with brain iron accumulation 8, 617917
CTH	140.6	100.0%	99.7%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTNS	118.7	100.0%	99.6%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTSA	146.1	100.0%	100.0%	Galactosialidosis, 256540
CTSC	119.4	100.0%	100.0%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010

CTSD	187.3	100.0%	99.0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	90.0	100.0%	100.0%	Pycnodysostosis, 265800
CUBN	103.2	99.6%	98.0%	Megaloblastic anemia-1, Finnish type, 261100
CYB561	160.2	92.8%	92.8%	Orthostatic hypotension 2, 618182
CYB5R3	163.2	99.6%	98.5%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	130.1	99.2%	94.8%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	171.1	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	170.2	100.0%	100.0%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Aldosterone to renin ratio raised, 0
CYP17A1	116.2	100.0%	99.8%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	128.2	99.7%	97.7%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	153.6	100.0%	100.0%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	102.4	99.9%	97.2%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	184.4	100.0%	99.8%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	164.1	100.0%	99.8%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	133.4	99.9%	97.5%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	139.8	99.1%	96.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	103.2	99.7%	97.2%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	157.7	100.0%	99.8%	D-2-hydroxyglutaric aciduria, 600721
DAO	110.4	100.0%	99.6%	No OMIM Disease ID
DBH	161.3	100.0%	99.9%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	109.6	99.7%	96.9%	Maple syrup urine disease, type II, 248600
DCXR	189.3	100.0%	100.0%	No OMIM disease ID
DDC	100.3	99.4%	96.2%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	166.9	100.0%	99.1%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	123.1	100.0%	99.9%	?Congenital disorder of glycosylation, type Ir, 614507
DEGS1	150.2	100.0%	100.0%	Leukodystrophy, hypomyelinating, 18, 618404
DGAT1	167.3	97.0%	92.8%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	132.2	100.0%	98.3%	Nephrotic syndrome, type 7, 615008

DGUOK	127.0	100.0%	98.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	170.7	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	84.5	97.3%	94.0%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DHFR	48.6	92.6%	80.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	107.2	100.0%	100.0%	Miller syndrome, 263750
DLD	117.2	100.0%	99.9%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	135.4	100.0%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	144.7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	92.5	99.1%	90.4%	3-methylglutaconic aciduria, type V, 610198
DNM1L	120.8	99.9%	97.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	134.2	99.8%	97.7%	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMT1	122.3	99.4%	98.7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	125.5	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	171.4	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	93.2	100.0%	100.0%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	134.2	95.5%	87.7%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	95.4	100.0%	99.1%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	218.6	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPYD	140.7	99.4%	96.2%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	121.4	100.0%	99.9%	Dihydropyrimidinuria, 222748
DTYMK	116.0	100.0%	100.0%	No OMIM Disease ID
EBP	68.9	99.8%	96.3%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	111.6	100.0%	100.0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL1	90.9	99.7%	96.6%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527

ELOVL4	103.3	100.0%	99.6%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	186.3	100.0%	100.0%	?Glycogen storage disease XIII, 612932
EOGT	103.3	79.5%	78.1%	Adams-Oliver syndrome 4, 615297
EPHX1	124.0	99.2%	96.3%	?Hypercholanemia, familial, 607748
EPHX2	100.4	99.7%	97.8%	No OMIM Disease ID
ETFA	132.0	100.0%	99.8%	Glutaric acidemia IIA, 231680
ETFB	127.7	100.0%	100.0%	Glutaric acidemia IIB, 231680
ETFDH	112.7	100.0%	99.7%	Glutaric acidemia IIC, 231680
ETHE1	105.9	99.9%	97.9%	Ethylmalonic encephalopathy, 602473
EXT1	91.1	99.9%	98.4%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	120.9	99.9%	99.0%	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682
FA2H	101.5	99.3%	95.1%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	136.7	100.0%	99.8%	Tyrosinemia, type I, 276700
FAR1	72.4	97.6%	91.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBP1	114.0	100.0%	99.2%	Fructose-1,6-bisphosphatase deficiency, 229700
FDFT1	150.3	100.0%	99.8%	Squalene synthase deficiency, 618156
FECH	107.9	100.0%	99.6%	Protoporphyria, erythropoietic, 1, 177000
FH	126.0	95.9%	89.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	178.0	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	108.0	99.9%	96.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLAD1	184.5	100.0%	99.8%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	133.3	99.9%	98.5%	Trimethylaminuria, 602079
FOLR1	115.7	100.0%	100.0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	129.8	99.1%	96.1%	Glutamate formiminotransferase deficiency, 229100
FUCA1	135.9	100.0%	100.0%	Fucosidosis, 230000
FUK	116.4	99.6%	98.4%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FUT2	163.2	100.0%	100.0%	No OMIM Disease ID

FUT6	159.8	100.0%	100.0%	Fucosyltransferase 6 deficiency, 613852
G6PC	149.8	100.0%	100.0%	Glycogen storage disease Ia, 232200
G6PC3	126.1	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	126.2	99.8%	98.4%	Hemolytic anemia, G6PD deficient (favism), 300908
GAA	180.3	100.0%	100.0%	Glycogen storage disease II, 232300
GAD1	114.6	100.0%	99.8%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	103.0	99.8%	98.1%	Krabbe disease, 245200
GALE	153.0	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALK1	186.1	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALM	93.1	100.0%	99.9%	No OMIM Disease ID
GALNS	118.1	100.0%	99.4%	Mucopolysaccharidosis IVA, 253000
GALNT3	126.0	99.9%	98.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	165.3	100.0%	100.0%	Galactosemia, 230400
GAMT	125.7	99.7%	94.3%	Cerebral creatine deficiency syndrome 2, 612736
GANAB	113.2	100.0%	98.9%	Polycystic kidney disease 3, 600666
GATM	139.0	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GBA	180.2	100.0%	100.0%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GBA2	151.6	100.0%	99.9%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GCDH	158.7	100.0%	99.7%	Glutaricaciduria, type I, 231670
GCH1	91.0	99.9%	99.4%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCK	152.9	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 MODY, type II, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCLC	148.2	99.6%	97.0%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCLM	101.7	99.7%	96.3%	No OMIM Disease ID
GCSH	32.0	94.3%	74.1%	?Glycine encephalopathy, 605899
GFPT1	142.7	99.9%	99.3%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GIF	110.0	100.0%	99.4%	Intrinsic factor deficiency, 261000

GK	43.3	82.3%	61.8%	Glycerol kinase deficiency, 307030
GLA	74.4	99.4%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	87.4	99.5%	95.2%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	60.8	91.8%	80.4%	Glycine encephalopathy, 605899
GLRA1	103.2	100.0%	99.8%	Hyperekplexia 1, 149400
GLRX5	149.3	99.8%	97.8%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	66.3	98.0%	88.9%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	80.0	99.8%	97.3%	Glutamine deficiency, congenital, 610015
GLYCTK	175.0	100.0%	99.8%	D-glyceric aciduria, 220120
GM2A	129.1	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMPPA	158.4	100.0%	100.0%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	233.1	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	118.6	98.2%	94.3%	No OMIM Disease ID
GNE	115.7	100.0%	99.3%	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	135.6	99.9%	98.8%	Glycine N-methyltransferase deficiency, 606664
GNPAT	128.8	99.7%	96.6%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	149.5	100.0%	99.4%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	199.0	99.9%	99.4%	Mucolipidosis III gamma, 252605
GNS	93.6	99.9%	97.2%	Mucopolysaccharidosis type IIID, 252940
GOT1	111.8	100.0%	99.5%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	85.1	96.6%	91.6%	No OMIM Disease ID
GPD1	94.7	100.0%	99.6%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	132.4	100.0%	99.9%	Brugada syndrome 2, 611777
GPHN	147.8	99.8%	98.8%	Molybdenum cofactor deficiency C, 615501
GPI	152.9	100.0%	100.0%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPT2	130.4	100.0%	99.6%	Mental retardation, autosomal recessive 49, 616281
GPX1	57.9	99.9%	97.6%	No OMIM Disease ID
GRHPR	106.8	85.1%	82.3%	Hyperoxaluria, primary, type II, 260000

GSS	98.9	100.0%	99.6%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GUSB	106.6	92.6%	91.1%	Mucopolysaccharidosis VII, 253220
GYG1	126.6	100.0%	99.8%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	116.9	100.0%	99.2%	Glycogen storage disease 0, muscle, 611556
GYS2	119.6	99.8%	97.5%	Glycogen storage disease 0, liver, 240600
H6PD	219.1	99.0%	99.0%	Cortisone reductase deficiency 1, 604931
HADH	118.2	99.3%	99.2%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	74.6	96.1%	89.6%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	76.9	96.0%	83.7%	Trifunctional protein deficiency, 609015
HAGH	154.4	100.0%	100.0%	No OMIM Disease ID
HEXA	112.3	93.8%	92.6%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	173.2	99.8%	97.3%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	114.7	100.0%	99.1%	Hemochromatosis, 235200
HGD	101.4	100.0%	99.7%	Alkaptonuria, 203500
HGSNAT	99.9	88.2%	86.3%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	103.8	98.2%	92.9%	No OMIM Disease ID
HIBCH	69.7	95.5%	75.9%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	123.7	100.0%	99.6%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HLCS	148.0	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMBS	102.8	100.0%	99.0%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	124.9	100.0%	99.5%	HMG-CoA lyase deficiency, 246450
HMGCS2	107.6	100.0%	99.6%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	153.8	98.0%	91.0%	Heme oxygenase-1 deficiency, 614034
HNF1A	179.0	100.0%	100.0%	MODY, type III, 600496 Hepatic adenoma, somatic, 142330

				Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HNF4A	140.7	100.0%	99.3%	MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HOGA1	163.0	100.0%	99.5%	Hyperoxaluria, primary, type III, 613616
HPD	159.7	100.0%	99.9%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	56.9	97.8%	87.8%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	81.7	99.3%	95.3%	No OMIM Disease ID
HSD11B1	116.2	100.0%	99.3%	Cortisone reductase deficiency 2, 614662
HSD11B2	183.2	95.8%	89.4%	Apparent mineralocorticoid excess, 218030
HSD17B10	98.0	100.0%	99.5%	HSD10 mitochondrial disease, 300438
HSD17B3	119.0	100.0%	100.0%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	106.4	95.5%	93.1%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	137.1	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	157.0	99.8%	97.4%	Bile acid synthesis defect, congenital, 1, 607765
HTRA2	145.3	100.0%	99.7%	3-methylglutaconic aciduria, type VIII, 617248
HYAL1	121.3	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492
IDH2	107.4	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	136.5	95.8%	95.4%	Retinitis pigmentosa 46, 612572
IDI1	60.9	99.6%	96.3%	No OMIM Disease ID
IDS	105.1	99.8%	97.2%	Mucopolysaccharidosis II, 309900
IDUA	169.2	99.3%	96.4%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IMPAD1	181.6	100.0%	100.0%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	57.4	97.6%	87.3%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
INPP5E	131.1	100.0%	99.3%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPPL1	139.7	99.9%	98.9%	Opsismodysplasia, 258480
INSR	123.5	99.4%	96.1%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200

IREB2	129.0	100.0%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
ISPD	112.0	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITCH	115.3	95.5%	94.5%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITPA	142.5	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647
IVD	106.7	100.0%	100.0%	Isovaleric acidemia, 243500
KCNA2	132.6	100.0%	99.4%	Epileptic encephalopathy, early infantile, 32, 616366
KCNJ11	222.1	100.0%	100.0%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KMT2A	138.5	100.0%	99.9%	Wiedemann-Steiner syndrome, 605130
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
L2HGDH	123.6	99.2%	97.2%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	89.8	97.8%	92.3%	Danon disease, 300257
LARGE1	122.8	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LCAT	156.5	99.6%	96.1%	Norum disease, 245900 Fish-eye disease, 136120
LCT	128.1	99.9%	98.2%	Lactase deficiency, congenital, 223000
LDHA	57.3	97.7%	89.0%	Glycogen storage disease XI, 612933
LDHB	84.2	94.4%	82.2%	No OMIM Disease ID
LFNG	132.2	95.1%	89.4%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	124.4	100.0%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	107.3	96.6%	94.9%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	104.4	100.0%	99.8%	Hepatic lipase deficiency, 614025
LIPT1	199.3	100.0%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	107.4	100.0%	99.9%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	95.5	97.1%	91.2%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LPIN1	128.5	99.5%	97.0%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	101.1	100.0%	99.7%	Majeed syndrome, 609628
LPL	133.6	100.0%	100.0%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LRAT	252.8	100.0%	100.0%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341

LTC4S	96.2	94.6%	79.7%	No OMIM Disease ID
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
MAN1B1	137.5	100.0%	99.9%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	139.1	99.9%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MANBA	117.1	99.7%	98.1%	Mannosidosis, beta, 248510
MAOA	100.1	100.0%	99.2%	Brunner syndrome, 300615
MAT1A	154.2	99.7%	98.2%	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MBOAT7	121.9	100.0%	99.9%	Mental retardation, autosomal recessive 57, 617188
MCCC1	138.0	100.0%	99.6%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	121.8	100.0%	99.9%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	124.7	100.0%	100.0%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	170.5	100.0%	99.4%	Mucolipidosis IV, 252650
MDH1	103.8	99.9%	99.0%	No OMIM Disease ID
MFSD2A	121.3	100.0%	99.6%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	117.4	100.0%	99.6%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	155.6	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	175.7	100.0%	99.7%	No OMIM Disease ID
MLYCD	105.5	99.7%	97.3%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168.7	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.3	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	214.4	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	76.8	93.0%	77.2%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOCOS	156.5	100.0%	99.2%	Xanthinuria, type II, 603592
MOCS1	101.3	99.3%	96.6%	Molybdenum cofactor deficiency A, 252150
MOCS2	134.2	99.6%	99.6%	Molybdenum cofactor deficiency B, 252160
MOGS	157.9	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	110.1	100.0%	99.8%	Congenital disorder of glycosylation, type If, 609180
MPI	115.8	100.0%	99.9%	Congenital disorder of glycosylation, type Ib, 602579
MRPL44	130.4	100.0%	99.9%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS36	66.8	91.7%	67.3%	No OMIM Disease ID
MSMO1	47.0	96.2%	87.7%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834

MTHFD1	119.4	99.9%	98.3%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	124.2	98.5%	96.7%	Homocystinuria due to MTHFR deficiency, 236250
MTM1	78.4	98.4%	91.8%	Myotubular myopathy, X-linked, 310400
MTMR2	99.0	99.9%	98.2%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	134.7	100.0%	99.6%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	135.6	100.0%	99.2%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MUT	128.8	100.0%	99.0%	Methylmalonic aciduria, mut(0) type, 251000
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
NADK2	163.3	100.0%	99.8%	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	131.4	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	130.5	98.5%	95.6%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	118.8	100.0%	100.0%	N-acetylglutamate synthase deficiency, 237310
NANS	105.0	100.0%	99.3%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEU1	150.1	99.5%	96.5%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NGLY1	134.1	100.0%	99.9%	Congenital disorder of deglycosylation, 615273
NMNAT1	118.9	99.9%	98.3%	Leber congenital amaurosis 9, 608553
NNT	127.0	99.9%	98.1%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NPC1	120.3	100.0%	99.4%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	130.7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPL	114.9	100.0%	99.7%	No OMIM Disease ID
NSD1	152.6	100.0%	99.8%	Sotos syndrome 1, 117550
NSDHL	133.7	99.9%	98.2%	CHILD syndrome, 308050 CK syndrome, 300831
NT5C3A	62.1	97.2%	83.5%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	157.1	100.0%	99.9%	Calcification of joints and arteries, 211800
NUS1	51.9	72.8%	44.9%	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	69.1	80.2%	69.8%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870

OCRL	106.2	99.9%	98.6%	Lowe syndrome, 309000 Dent disease 2, 300555
OPA3	171.9	100.0%	99.9%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPLAH	158.6	100.0%	100.0%	5-oxoprolinase deficiency, 260005
OTC	111.3	100.0%	99.7%	Ornithine transcarbamylase deficiency, 311250
OXCT1	123.6	99.7%	98.2%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	128.9	100.0%	100.0%	Phenylketonuria, 261600
PANK2	161.5	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	170.2	99.9%	98.8%	Pyruvate carboxylase deficiency, 266150
PCBD1	109.5	100.0%	99.8%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	97.7	99.1%	95.4%	Propionicacidemia, 606054
PCCB	114.9	99.5%	97.1%	Propionicacidemia, 606054
PCK1	128.7	100.0%	100.0%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	177.6	100.0%	100.0%	No OMIM disease ID
PCYT1A	97.1	99.1%	95.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PCYT2	159.4	100.0%	99.7%	No OMIM Disease ID
PDSS1	106.9	97.6%	88.2%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	115.4	99.3%	95.2%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	126.5	100.0%	99.9%	Prolidase deficiency, 170100
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	123.8	100.0%	98.4%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	93.3	100.0%	99.9%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	125.4	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	189.6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	144.7	99.8%	98.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	157.0	98.9%	95.7%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	85.8	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	137.4	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867

PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.6	100.0%	99.6%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	119.4	99.9%	99.3%	Glycogen storage disease VII, 232800
PGAM2	180.7	100.0%	100.0%	Glycogen storage disease X, 261670
PGAP1	106.2	98.8%	94.3%	Mental retardation, autosomal recessive 42, 615802
PGAP2	145.1	100.0%	99.8%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	74.5	63.7%	60.6%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	47.0	92.1%	78.7%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	134.8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PGM3	148.4	100.0%	99.9%	Immunodeficiency 23, 615816
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	90.2	97.8%	91.7%	Muscle glycogenosis, 300559
PHKA2	97.2	100.0%	99.2%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	124.0	100.0%	99.2%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	123.2	99.9%	98.4%	No OMIM Disease ID
PHKG2	171.3	100.0%	100.0%	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency, 0
PHYH	75.9	100.0%	97.9%	Refsum disease, 266500
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	91.5	99.9%	95.9%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGL	132.2	100.0%	99.1%	CHIME syndrome, 280000
PIGM	157.7	100.0%	100.0%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	103.7	93.6%	90.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	157.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 2, 614749

PIGP	88.3	95.6%	86.3%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGQ	151.8	94.8%	92.6%	Epileptic encephalopathy, early infantile, 77, 618548
PIGT	169.4	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	129.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	144.9	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	89.9	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	122.5	100.0%	99.8%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PIK3R1	125.7	99.9%	98.8%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	115.8	95.4%	91.8%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	130.3	100.0%	100.0%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	136.7	99.9%	99.3%	Corneal fleck dystrophy, 121850
PIP5K1C	151.5	99.9%	98.4%	Lethal congenital contractural syndrome 3, 611369
PKLR	189.0	100.0%	99.9%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PLA2G5	111.2	100.0%	100.0%	No OMIM disease ID
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLA2G7	117.7	100.0%	99.1%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLCB1	134.5	100.0%	99.8%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	102.7	99.8%	97.9%	Auriculocondylar syndrome 2, 614669
PLCD1	127.9	100.0%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	129.1	99.9%	99.2%	Nephrotic syndrome, type 3, 610725

PLCG2	110.7	100.0%	99.5%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLIN1	107.2	100.0%	99.8%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	141.5	99.9%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	115.6	99.4%	96.2%	Bruck syndrome 2, 609220
PLOD3	120.2	100.0%	99.8%	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	97.2	99.9%	97.2%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	135.2	99.9%	97.6%	?Pancreatic lipase deficiency, 614338
PNMT	119.6	100.0%	100.0%	No OMIM Disease ID
PNP	113.1	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	159.9	100.0%	99.9%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	153.1	100.0%	99.6%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
PNPO	78.3	100.0%	99.2%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	146.2	100.0%	99.2%	Dowling-Degos disease 2, 615327
POGLUT1	100.2	100.0%	99.1%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POLR3A	119.8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	132.0	99.9%	98.3%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	123.6	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	225.4	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	144.2	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	137.5	99.6%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	109.7	100.0%	99.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156

PPCS	166.9	100.0%	99.2%	Cardiomyopathy, dilated, 2C, 618189
PPM1K	139.2	100.0%	100.0%	?Maple syrup urine disease, mild variant, 615135
PPOX	101.7	99.9%	97.6%	Porphyria variegata, 176200
PPT1	140.2	90.3%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	135.6	99.1%	96.5%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKCSH	165.4	99.8%	96.3%	Polycystic liver disease 1, 174050
PRODH	88.9	91.8%	83.0%	Hyperprolinemia, type I, 239500
PRPS1	113.2	100.0%	99.9%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PSAP	103.3	100.0%	99.5%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	46.2	91.6%	74.2%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSPH	122.1	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PTEN	125.3	99.7%	95.5%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTGIS	123.3	99.8%	97.4%	Hypertension, essential, 145500
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTS	103.1	100.0%	98.3%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	105.0	100.0%	99.0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	129.0	99.7%	97.6%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	147.0	100.0%	99.9%	Glycogen storage disease VI, 232700
PYGM	130.4	100.0%	100.0%	McArdle disease, 232600
QDPR	103.7	99.9%	99.1%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RBCK1	118.4	100.0%	99.4%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895

RDH12	86.4	99.8%	98.1%	Leber congenital amaurosis 13, 612712
RDH5	182.9	100.0%	100.0%	Fundus albipunctatus, 136880
RFT1	106.6	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015
RINT1	158.2	99.8%	98.2%	Infantile liver failure syndrome 3, 618641
RPE65	133.0	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	119.9	100.0%	99.4%	Ribose 5-phosphate isomerase deficiency, 608611
SARDH	141.5	93.2%	91.6%	No OMIM disease ID
SAT1	129.2	100.0%	99.1%	No OMIM Disease ID
SC5D	149.4	100.0%	99.6%	Lathosterolosis, 607330
SCARB2	106.4	99.9%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	107.1	99.8%	96.8%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	161.4	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	132.1	99.7%	98.2%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SELENBP1	132.1	100.0%	99.9%	Extraoral halitosis due to MTO deficiency, 618148
SEPSECS	160.6	100.0%	100.0%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	110.4	100.0%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	152.5	98.1%	94.9%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SI	112.1	99.4%	95.3%	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	110.2	99.9%	99.1%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC12A1	144.9	100.0%	99.7%	Bartter syndrome, type 1, 601678
SLC13A3	94.6	100.0%	99.8%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC16A1	140.0	100.0%	99.3%	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC17A5	136.7	98.7%	95.1%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC18A2	112.3	100.0%	99.9%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC22A12	131.0	100.0%	99.9%	Hypouricemia, renal, 220150
SLC22A5	144.8	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140
SLC25A1	114.2	99.8%	97.0%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	118.2	99.9%	98.1%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	152.1	98.4%	94.4%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970

SLC25A19	82.0	100.0%	98.5%	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A20	96.7	100.0%	99.9%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	121.5	100.0%	99.9%	No OMIM Disease ID
SLC25A32	128.5	100.0%	100.0%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	98.5	99.0%	95.5%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	143.7	100.0%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC28A1	138.6	100.0%	99.9%	No OMIM disease ID
SLC2A1	160.0	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A2	159.5	100.0%	99.9%	Fanconi-Bickel syndrome, 227810
SLC2A9	108.3	100.0%	98.9%	Hypouricemia, renal, 2, 612076
SLC30A10	200.7	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	135.7	99.8%	97.0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	123.5	100.0%	99.8%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	114.2	100.0%	99.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	63.5	80.4%	77.0%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	209.1	100.0%	99.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	126.1	99.6%	95.7%	Schneckenbecken dysplasia, 269250
SLC37A4	122.0	100.0%	99.7%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC39A14	101.7	99.9%	98.8%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	130.4	100.0%	99.7%	Acrodermatitis enteropathica, 201100
SLC39A8	144.7	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	147.7	100.0%	99.7%	Cystinuria, 220100
SLC46A1	121.7	100.0%	98.0%	Folate malabsorption, hereditary, 229050
SLC52A1	219.5	100.0%	100.0%	Riboflavin deficiency, 615026
SLC52A2	213.2	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	128.9	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC5A1	116.9	100.0%	99.5%	Glucose/galactose malabsorption, 606824
SLC5A2	150.5	100.0%	100.0%	Renal glucosuria, 233100

SLC6A19	139.1	100.0%	100.0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A8	58.5	97.6%	87.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	110.7	100.0%	99.8%	Lysinuric protein intolerance, 222700
SLC7A9	126.6	100.0%	99.3%	Cystinuria, 220100
SLCO1B1	47.9	97.4%	88.7%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	47.0	96.7%	86.6%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	161.8	100.0%	99.6%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	63.5	88.9%	74.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNX14	79.9	99.7%	93.7%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOD1	129.8	100.0%	99.7%	Amyotrophic lateral sclerosis 1, 105400 Spastic tetraplegia and axial hypotonia, progressive, 618598
SOD2	208.3	100.0%	100.0%	No OMIM disease ID
SPR	159.7	100.0%	100.0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	108.0	98.4%	91.8%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	146.3	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQOR	103.2	99.9%	97.9%	No OMIM Disease ID
SRD5A2	92.3	100.0%	97.6%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	149.2	99.9%	98.5%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SSR4	118.5	100.0%	100.0%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	143.4	100.0%	99.8%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	104.4	89.3%	85.5%	Salt and pepper developmental regression syndrome, 609056
STAR	146.4	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
STS	81.1	99.6%	96.3%	Ichthyosis, X-linked, 308100
STT3A	125.4	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127.0	100.0%	99.8%	?Congenital disorder of glycosylation, type Ix, 615597
SUCLA2	57.8	91.5%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	104.2	100.0%	99.7%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	57.6	92.1%	79.9%	No OMIM Disease ID
SUGCT	126.9	99.0%	94.8%	Glutaric aciduria III, 231690
SUMF1	91.7	99.9%	97.6%	Multiple sulfatase deficiency, 272200
SUOX	180.8	100.0%	100.0%	Sulfite oxidase deficiency, 272300
TALDO1	158.9	100.0%	99.8%	Transaldolase deficiency, 606003

TANGO2	139.6	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	119.7	100.0%	99.9%	Tyrosinemia, type II, 276600
TAZ	125.3	99.3%	96.2%	Barth syndrome, 302060
TBXAS1	135.5	100.0%	100.0%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	157.6	100.0%	100.0%	Transcobalamin II deficiency, 275350
TECR	139.6	100.0%	99.9%	Mental retardation, autosomal recessive 14, 614020
TH	106.8	100.0%	99.2%	Segawa syndrome, recessive, 605407
TIMM50	133.8	100.0%	99.4%	3-methylglutaconic aciduria, type IX, 617698
TK2	111.5	100.0%	99.8%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKT	124.8	98.7%	98.3%	Short stature, developmental delay, and congenital heart defects, 617044
TMEM165	159.2	99.9%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	127.8	100.0%	99.9%	Congenital disorder of glycosylation, type IIp, 616829
TMEM5	167.7	99.8%	96.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM70	117.3	99.9%	98.5%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	87.6	99.9%	96.4%	No OMIM disease ID
TPI1	120.2	99.8%	97.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	96.3	99.7%	97.1%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPMT	40.3	96.1%	81.4%	No OMIM disease ID
TPP1	130.2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TRAK1	159.8	100.0%	99.7%	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	124.3	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC2L	211.9	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TREH	153.4	98.4%	93.9%	Trehalase deficiency, 612119
TUSC3	155.7	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TYMP	138.6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	150.8	100.0%	100.0%	No OMIM Disease ID
TYR	153.5	100.0%	100.0%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
TYRP1	155.1	100.0%	99.9%	Albinism, oculocutaneous, type III, 203290
UGT1A1	192.9	100.0%	100.0%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785

UMPS	156.7	100.0%	98.8%	Orotic aciduria, 258900
UPB1	150.9	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161
UROC1	143.4	100.0%	99.9%	?Urocanase deficiency, 276880
UROD	139.7	99.6%	96.7%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	104.6	100.0%	99.9%	Porphyria, congenital erythropoietic, 263700
VPS13B	135.9	99.4%	97.8%	Cohen syndrome, 216550
VPS33A	106.3	96.6%	94.7%	Mucopolysaccharidosis-plus syndrome, 617303
XDH	98.4	100.0%	99.8%	Xanthinuria, type I, 278300
XYLT1	138.1	100.0%	99.4%	Desbuquois dysplasia 2, 615777
XYLT2	161.8	99.9%	98.7%	Spondyloocular syndrome, 605822

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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors