METABOLIC DISORDERS GENE PANEL DG 2.14 (625 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AASS	128.8	99.6	97.4	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	92.7	100	99.5	GABA-transaminase deficiency, 613163
ABCD1	76	74.7	68	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	186.3	100	99.8	No OMIM phenotype
ABCD3	93.7	95.2	89.5	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	143.6	99.9	98.3	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	145.2	100	99.2	Sitosterolemia, 210250
ABCG8	148.4	99.2	96.6	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	107	97.3	88	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	209.6	100	99.9	Chanarin-Dorfman syndrome, 275630
ACACA	135.6	98.2	97.5	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	141.5	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADM	101.3	98.8	95.6	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	123.9	99.3	97.6	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	119.1	99.5	95.6	2-methylbutyrylglycinuria, 610006
ACADVL	119	98.7	95.1	VLCAD deficiency, 201475
ACAT1	123.7	99.2	94.6	Alpha-methylacetoacetic aciduria, 203750
ACAT2	159	100	100	?ACAT2 deficiency, 614055
ACBD5	145.3	97.8	96	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACO2	129.3	95.8	91.8	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	155.3	100	100	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	136.2	100	99.8	Bile acid synthesis defect, congenital, 6, 617308

ACSF3	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265
ACSL4	104.7	97.5	91.8	Mental retardation, X-linked 63, 300387
ACY1	132.8	99.9	98.3	Aminoacylase 1 deficiency, 609924
ADA	113	98.9	97.3	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	106.7	99.8	97.9	No OMIM phenotype
ADCY5	129.2	92.4	89.2	Dyskinesia, familial, with facial myokymia, 606703
ADK	100.4	99.5	96.1	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	183.6	99.2	99.1	Adenylosuccinase deficiency, 103050
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGK	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	51.7	96.8	84.8	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	139.5	99.9	99.2	Hyperoxaluria, primary, type 1, 259900
AHCY	124.5	100	99.8	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	119.8	100	99.2	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	111.8	99.8	96.6	Reticular dysgenesis, 267500
AKR1D1	106.1	98.5	94.3	Bile acid synthesis defect, congenital, 2, 235555
ALAD	100.6	99.8	97.4	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	89.7	99.6	97.1	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	131.1	100	99.9	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	104.7	93.4	89.6	Microphthalmia, isolated 8, 615113
ALDH2	124.7	100	99.7	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}, 0

ALDH3A2	125.7	95.3	94.6	Sjogren-Larsson syndrome, 270200
ALDH4A1	116	100	98.6	Hyperprolinemia, type II, 239510
ALDH5A1	87.6	86.4	80.1	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	127.3	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	77.1	93.7	85.4	Epilepsy, pyridoxine-dependent, 266100
ALDOA	139.1	76.3	74.7	Glycogen storage disease XII, 611881
ALDOB	165.7	100	99.4	Fructose intolerance, hereditary, 229600
ALG1	50.9	53.6	48.8	Congenital disorder of glycosylation, type Ik, 608540
ALG10	304.9	100	100	{Long QT syndrome, acquired, reduced susceptibility to}, 613688
ALG11	139.6	96.7	96	Congenital disorder of glycosylation, type Ip, 613661
ALG12	156.2	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	86.7	98.7	94.1	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG14	233.8	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	115.9	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.4	96	93.3	Congenital disorder of glycosylation, type Ic, 603147
ALG8	126	96.5	95.1	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	124.3	100	99.6	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALOX12B	130.6	100	99.5	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	156.4	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMACR	157.9	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	66.8	83.5	71.6	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	126.7	100	99.9	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	131.9	99.8	98.6	[AMP deaminase deficiency, erythrocytic], 612874
AMT	173.1	100	100	Glycine encephalopathy, 605899
AP1S1	111.3	99.9	99.5	MEDNIK syndrome, 609313

APOC2	99.4	100	100	Hyperlipoproteinemia, type Ib, 207750
APRT	68.2	100	98.7	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	167.8	100	100	Argininemia, 207800
ARSA	97.8	100	99.7	Metachromatic leukodystrophy, 250100
ARSB	117.5	94.9	87.7	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	105.9	97.6	92.1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	114.4	99.9	98.8	Argininosuccinic aciduria, 207900
ASPA	127.6	99.1	95.8	Canavan disease, 271900
ASS1	97.9	95.7	87.5	Citrullinemia, 215700
ATIC	119.5	99.7	99	AICA-ribosiduria due to ATIC deficiency, 608688
ATP6AP1	112.8	99.1	94.7	Immunodeficiency 47, 300972
ATP6V0A2	130	100	99.3	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V1A	144.8	98.2	94.9	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1E1	67.2	92.2	85.6	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	133.2	99.7	97.8	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	168.9	100	99.8	Wilson disease, 277900
ATP8B1	139	96.7	94.4	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
AUH	90.9	99.9	97.6	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	130.7	100	99.9	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	115	92.4	89.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	47.5	76.4	71.7	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	93.6	99.4	95.9	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	101.2	97.4	93.4	Peters-plus syndrome, 261540
B4GALT1	105.4	99.9	99	Congenital disorder of glycosylation, type IId, 607091

B4GALT7	104.3	96.1	95	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	120.4	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	121.4	98.3	95.3	Hypercholanemia, familial, 607748
BCKDHA	171.5	100	99.5	Maple syrup urine disease, type Ia, 248600
BCKDHB	112.6	88.9	81.3	Maple syrup urine disease, type Ib, 248600
BCO1	164.9	100	100	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	125.1	100	99.7	Hyperbiliverdinemia, 614156
BMP2	173.4	100	99.9	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BPGM	130.8	100	100	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BTD	166.6	100	99.9	Biotinidase deficiency, 253260
C1GALT1C1	147.2	99.8	98.7	Tn polyagglutination syndrome, somatic, 300622
CA5A	124.1	99.5	94.9	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	158.9	100	99.7	Epileptic encephalopathy, early infantile, 50, 616457
CANT1	142.1	100	99.8	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAT	148.5	100	100	Acatalasemia, 614097
CBS	116.2	97.1	91.1	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCDC115	59.3	88	85.3	Congenital disorder of glycosylation, type IIo, 616828
CEL	130	85	82.7	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	100.4	98.6	92.8	Retinitis pigmentosa 26, 608380
CERS3	106.8	100	98.8	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	124	99.1	96.3	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	117.5	99.6	97.3	[Chitotriosidase deficiency], 614122
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	165.6	95.7	93.3	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	91.6	100	97.5	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095

CHST6	334	100	100	Macular corneal dystrophy, 217800
CHSY1	138.4	95.9	93.9	Temtamy preaxial brachydactyly syndrome, 605282
CLN3	114.9	92.5	90.7	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.1	98.2	92.2	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.6	98.9	95.3	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	163.9	83.5	83.5	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COG1	124.2	100	99.9	Congenital disorder of glycosylation, type IIg, 611209
COG2	122.6	97.2	94.7	?Congenital disorder of glycosylation, type IIq, 617395
COG4	123.8	100	99.9	Congenital disorder of glycosylation, type IIj, 613489
COG5	107	97.4	93.8	Congenital disorder of glycosylation, type IIi, 613612
COG6	78.4	95	85.9	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	125.1	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	122.4	99.9	98.4	Congenital disorder of glycosylation, type IIh, 611182
COMT	206.6	100	99.9	{Panic disorder, susceptibility to}, 167870 {Schizophrenia, susceptibility to}, 181500
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88.4	84.9	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	184.4	100	100	No OMIM phenotype Cerebellar ataxia and static encephalopmyopathy (Malicdan (2018) Hum Mutat 39,69) Intellectual disability (Najmabadi (2011) Nature 478,57)
COQ6	143.9	99.3	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	158.5	99.7	98.9	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	134.3	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	90.5	100	99.1	Nephrotic syndrome, type 9, 615573
COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
CP	120	93.9	89.6	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPOX	116.8	95.2	88.1	Coproporphyria, 121300

				Harderoporphyria, 121300
CPS1	143.8	100	99.8	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	169.3	100	98.7	CPT deficiency, hepatic, type IA, 255120
CPT2	162.8	97.2	95.4	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	152.6	99.9	98.1	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTNS	120.1	100	99.9	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTSA	134.1	100	99.4	Galactosialidosis, 256540
CTSC	127.5	100	100	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	163.7	98	95.3	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	105	100	99.9	Pycnodysostosis, 265800
CUBN	127.8	99.8	98.4	Megaloblastic anemia-1, Finnish type, 261100
CYB561	131	92.8	92.6	No OMIM phenotype Orthostatic hypotension (van den Berg (2018) Circ Res 122,846)
CYB5R3	147.3	98	98	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	123.9	99.6	97.7	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	175.9	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	173.4	100	100	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	135	100	99.7	17,20-lyase deficiency, isolated, 202110

				17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	160.6	99.1	97.3	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	134.8	100	100	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP21A2	93.8	95.8	86.6	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	175.1	98.3	96.1	Cerebrotendinous xanthomatosis, 213700
CYP27B1	137.1	100	99.1	Vitamin D-dependent rickets, type I, 264700
CYP2R1	138.5	96.4	86.6	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	119.2	93.7	91.2	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	93.2	94.7	87.7	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	134.5	97.5	95.2	D-2-hydroxyglutaric aciduria, 600721
DAO	134.5	100	99.9	{Schizophrenia}, 181500
DBH	140.7	100	99.8	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma], 0
DBT	102.1	97.3	93.8	Maple syrup urine disease, type II, 248600
DCXR	163	99.9	98.4	[Pentosuria], 260800
DDC	101	99.1	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	141.8	97.1	94.8	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	120.5	99.9	99.1	?Congenital disorder of glycosylation, type Ir, 614507
DGAT1	156.2	88.5	86.3	?Diarrhea 7, 615863
DGKE	142.3	99.5	95.2	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	183	100	100	Desmosterolosis, 602398
DHCR7	158.3	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	93.5	97.8	94.8	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861

DHFR	48.4	91.1	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	92.2	100	99.9	Miller syndrome, 263750
DLD	123.5	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	157	98.8	97.2	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	129.3	87.4	87.3	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	97.9	98.5	90	3-methylglutaconic aciduria, type V, 610198
DNM1L	123.5	99.7	96.6	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	127.4	97.5	94.4	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	113.4	99.2	98.3	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	124.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.1	100	99.4	Congenital disorder of glycosylation, type Iu, 615042
DPM3	183.7	100	100	Congenital disorder of glycosylation, type Io, 612937
DPYD	158.3	95.6	93.7	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	133.5	100	99.5	Dihydropyrimidinuria, 222748
EBP	83.3	100	98	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECHS1	112.8	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL4	91.9	99.9	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ENO3	179.3	100	100	?Glycogen storage disease XIII, 612932
EOGT	119.9	79.5	78.4	Adams-Oliver syndrome 4, 615297
EPHX1	122.6	98.8	96.1	?Hypercholanemia, familial, 607748
EPHX2	114.2	100	99.1	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890

ETFA	143.3	100	99.4	Glutaric acidemia IIA, 231680
ETFB	126.6	100	100	Glutaric acidemia IIB, 231680
ETFDH	105.4	100	99.3	Glutaric acidemia IIC, 231680
ETHE1	85.5	99.3	95.8	Ethylmalonic encephalopathy, 602473
EXT1	105.4	99.9	98.5	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.5	99.9	99.1	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
FA2H	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319
FAH	151.3	100	99.9	Tyrosinemia, type I, 276700
FBP1	127	100	98.8	Fructose-1,6-bisphosphatase deficiency, 229700
FDFT1	153.6	98.1	95.7	No OMIM phenotype
FECH	121.9	99.9	99.4	Protoporphyria, erythropoietic, 1, 177000
FH	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	94.5	100	99.7	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	120.2	99.2	94.2	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	191.6	100	98.9	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	153.5	99.9	99.2	Trimethylaminuria, 602079
FOLR1	150.4	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	89.8	94.6	89.8	Glutamate formiminotransferase deficiency, 229100
FUCA1	135	100	99.5	Fucosidosis, 230000
FUT2	173.7	100	100	[Bombay phenotype], 0 {Norwalk virus infection, resistance to}, 0 {Vitamin B12 plasma level QTL1}, 612542
FUT6	165.8	100	100	Fucosyltransferase 6 deficiency, 613852
G6PC	180.7	100	100	Glycogen storage disease Ia, 232200
G6PC3	123.7	100	100	Dursun syndrome, 612541

				Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	118.3	99.5	97.5	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	128.5	100	99.9	Glycogen storage disease II, 232300
GAD1	128.7	99.9	98.4	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	100.6	98.9	94.6	Krabbe disease, 245200
GALE	154.8	100	100	Galactose epimerase deficiency, 230350
GALK1	125.4	100	99.7	Galactokinase deficiency with cataracts, 230200
GALNS	93.2	99	95.6	Mucopolysaccharidosis IVA, 253000
GALNT3	128.2	99.2	96	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	168.7	100	100	Galactosemia, 230400
GAMT	93.5	90.9	80.7	Cerebral creatine deficiency syndrome 2, 612736
GANAB	120.1	99.9	98.9	Polycystic kidney disease 3, 600666
GATM	150.6	100	100	Cerebral creatine deficiency syndrome 3, 612718
GBA	240.3	100	100	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	176.2	99.9	99.3	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	147.6	99.9	99.1	Glutaricaciduria, type I, 231670
GCH1	74.4	97	86.5	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	141.4	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	133.2	99.9	98.9	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	106	98	88.2	{Myocardial infarction, susceptibility to}, 608446

GCSH	34.2	83.1	67.8	?Glycine encephalopathy, 605899
GFPT1	144.4	99.9	97.6	Myasthenia, congenital, 12, with tubular aggregates, 610542
GIF	141.2	100	99.9	Intrinsic factor deficiency, 261000
GK	43.6	74	54.7	Glycerol kinase deficiency, 307030
GLA	81.3	99.7	97.6	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	78.9	90.6	82.7	Glycine encephalopathy, 605899
GLRA1	123.3	100	100	Hyperekplexia 1, 149400
GLRX5	108.2	92.6	83.8	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	74.5	94.4	84.3	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	108.8	99.9	98.2	Glutamine deficiency, congenital, 610015
GLYCTK	202.6	100	99.5	D-glyceric aciduria, 220120
GM2A	139.6	100	100	GM2-gangliosidosis, AB variant, 272750
GMPPA	136.8	100	99.9	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	228.5	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GMPS	112.9	96.9	89.2	No OMIM phenotype Leukemia, acute myelogenous, 601626
GNE	153.8	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	147.2	99.4	97	Glycine N-methyltransferase deficiency, 606664
GNPAT	133.6	99.4	96.4	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	151.6	96.1	89.7	Mucolipidosis III gamma, 252605
GNS	107.9	96.9	92	Mucopolysaccharidosis type IIID, 252940
GOT1	127	100	99.5	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	102.7	94.5	89.2	No OMIM phenotype

GPD1	93.5	99.9	99.1	Hypertriglyceridemia, transient infantile, 614480
GPD1L	138.4	100	98.5	Brugada syndrome 2, 611777
GPHN	167.2	98.4	96.9	Molybdenum cofactor deficiency C, 615501
GPI	142.6	100	99.5	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPT2	138	98.2	90.9	Mental retardation, autosomal recessive 49, 616281
GPX1	46.2	94.9	83.2	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GRHPR	112.5	85.1	78.2	Hyperoxaluria, primary, type II, 260000
GSS	104	100	99.8	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GUSB	116.1	92.2	89.4	Mucopolysaccharidosis VII, 253220
GYG1	157.8	100	99.6	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	110.8	100	98.5	Glycogen storage disease 0, muscle, 611556
GYS2	150.2	98.5	93.9	Glycogen storage disease 0, liver, 240600
H6PD	169.5	99	99	Cortisone reductase deficiency 1, 604931
HADH	110.8	98	95.1	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	84.4	96.5	90.3	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.5	92.5	79.5	Trifunctional protein deficiency, 609015
HAGH	130.5	99.8	97.8	[Glyoxalase II deficiency], 614033
HEXA	118.3	93.8	92.2	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	129.7	99.4	94	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	142.3	100	99.7	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	127.8	100	99.8	Alkaptonuria, 203500

HGSNAT	101	86.4	85.7	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	105.2	92.1	91	No OMIM phenotype
HIBCH	67.7	92.7	69.5	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	143.4	100	99.9	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HLCS	172.8	100	100	Holocarboxylase synthetase deficiency, 253270
HMBS	109	100	99.8	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	143.3	100	99.9	HMG-CoA lyase deficiency, 246450
HMGCS2	131.5	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	128.7	95.8	89.5	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HOGA1	147.5	99.8	98.1	Hyperoxaluria, primary, type III, 613616
HPD	137.8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPRT1	58.2	96	84.8	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	75.3	94.8	85	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD11B1	147.1	100	99.9	Cortisone reductase deficiency 2, 614662
HSD11B2	165.2	85.7	82.5	Apparent mineralocorticoid excess, 218030
HSD17B10	117.1	100	99.2	HSD10 mitochondrial disease, 300438
HSD17B3	156.4	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95.1	93.9	90.8	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	189.2	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	136.3	98.2	91	Bile acid synthesis defect, congenital, 1, 607765
HTRA2	122.1	100	99.7	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
HYAL1	115.3	100	100	?Mucopolysaccharidosis type IX, 601492
IDH2	103.5	99.6	96.9	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	165.5	95.9	95.4	Retinitis pigmentosa 46, 612572
IDS	111.3	99.6	98.3	Mucopolysaccharidosis II, 309900

IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IMPAD1	147.2	99.9	99.4	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	61.3	87.8	83.5	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
INPP5E	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	123.6	96.7	93.7	Opsismodysplasia, 258480
ISPD	104.4	95.2	84.8	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
IVD	114.9	100	100	Isovaleric acidemia, 243500
KMT2A	152.5	99.3	98.6	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	142.1	99.9	99	Kabuki syndrome 1, 147920
L2HGDH	129.1	98.4	97	L-2-hydroxyglutaric aciduria, 236792
LAMP2	106.1	92.7	91.2	Danon disease, 300257
LARGE1	143	100	99.6	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	145.6	97.8	91.7	Fish-eye disease, 136120 Norum disease, 245900
LCT	142	99.9	99.1	Lactase deficiency, congenital, 223000
LDHA	59.8	94.1	87.1	Glycogen storage disease XI, 612933
LDHB	106.8	97.3	85.6	[Lactate dehydrogenase-B deficiency], 614128
LFNG	91.6	85.4	83.3	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	133.7	99.5	97.1	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	110.9	98.8	95.8	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	115.2	100	99.8	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPT1	227.4	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	92.2	97.3	83.2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	80.2	91.9	83	Methylmalonic aciduria and homocystinuria, cblF type, 277380

LPIN1	134.6	99.8	97.8	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	111.5	100	99.6	Majeed syndrome, 609628
LPL	147.2	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LRAT	298.3	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	54.3	71.8	61.4	Leukotriene C4 synthase deficiency, 614037
LYST	134.6	97.8	93.9	Chediak-Higashi syndrome, 214500
MAN1B1	128.9	100	99.7	Mental retardation, autosomal recessive 15, 614202
MAN2B1	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAOA	113.8	99.9	98.7	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAT1A	185.4	99.7	97.5	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MCCC1	151.7	100	99.4	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	131.1	99.9	98.9	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	99.8	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	150.2	98.8	97	Mucolipidosis IV, 252650
MFSD2A	115.1	98.8	95.9	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	125.1	99.9	98.4	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	157.2	100	99.9	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	147.8	98.8	94.8	Thyroid carcinoma, follicular, 188470
MLYCD	75.8	91.2	86.8	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	183.2	100	99.6	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.2	100	99.9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	205.8	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	77	89.3	75	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410

				Methylmalonic aciduria, cblD type, variant 2, 277410
MOCS1	87.4	98.4	93	Molybdenum cofactor deficiency A, 252150
MOCS2	139.5	99.6	99.6	Molybdenum cofactor deficiency B, 252160
MOGS	121.6	99.8	99.1	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	111.8	100	99.7	Congenital disorder of glycosylation, type If, 609180
MPI	146.2	100	100	Congenital disorder of glycosylation, type Ib, 602579
MSMO1	45.8	92.6	78.5	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	139.6	99.8	98.4	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	126.1	98.4	97.2	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	93.7	99.2	93	Myotubular myopathy, X-linked, 310400
MTMR2	106.6	100	99.2	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	140.9	99.8	98.8	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	139.1	100	99.2	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUT	121.8	99.2	95.1	Methylmalonic aciduria, mut(0) type, 251000
MVK	124.3	92.1	90.4	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
NAGA	139.4	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	108.7	92.4	90.4	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	81	98	91.6	N-acetylglutamate synthase deficiency, 237310
NANS	106.1	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NEU1	148.1	99.4	97.1	Sialidosis, type I, 256550 Sialidosis, type II, 256550

NMNAT1	137.9	100	99.7	Leber congenital amaurosis 9, 608553
NNT	136.9	98.6	97.1	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NPC1	147.9	99.2	97.8	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	140.7	100	99.9	Niemann-pick disease, type C2, 607625
NSD1	155.2	100	99.9	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	169.2	99.9	98.2	CHILD syndrome, 308050 CK syndrome, 300831
NT5C3A	62	89.3	78.8	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	166.3	100	99.8	Calcification of joints and arteries, 211800
NUS1	69.6	62	40.7	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
OAT	89.2	77.7	70.5	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	122.2	98.8	96.3	Dent disease 2, 300555 Lowe syndrome, 309000
OPA3	128	99.5	97.4	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	109.4	99.4	97.9	5-oxoprolinase deficiency, 260005
OTC	123.3	99.9	99.4	Ornithine transcarbamylase deficiency, 311250
OXCT1	121.4	99.6	97.8	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	151.7	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	146.6	99.3	93.1	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	149.3	97.7	94.6	Pyruvate carboxylase deficiency, 266150
PCBD1	113.3	99.5	99.1	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	103.1	96.4	89.3	Propionicacidemia, 606054
PCCB	129.7	98.7	96.5	Propionicacidemia, 606054
PCK1	143	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	194.5	100	99.8	PEPCK deficiency, mitochondrial, 261650
PCYT1A	113.5	98.3	94.7	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDSS1	116.7	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652

PEPD	116	99.6	98.5	Prolidase deficiency, 170100
PEX1	115.8	97.7	95.4	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	111.8	96.1	90.1	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	105.7	99.7	98.3	?Peroxisome biogenesis disorder 14B, 614920
PEX12	168.3	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.6	99.8	98.7	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	149	99.7	97.5	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	137	97.1	93.1	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	92.9	99.9	99.2	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147.1	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	76.4	100	99.8	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	98.1	99.1	94.3	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	111.7	99.9	98.3	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	94.5	90.4	86.1	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	113.5	89.6	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	150.4	100	99.8	Glycogen storage disease VII, 232800
PGAM2	170.9	100	99.9	Glycogen storage disease X, 261670
PGAP1	98.1	94.9	88.6	Mental retardation, autosomal recessive 42, 615802
PGAP2	158.4	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	72.8	62.5	58	Hyperphosphatasia with mental retardation syndrome 4, 615716

PGK1	54.5	93.3	81.6	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	133.9	100	99.9	Congenital disorder of glycosylation, type It, 614921
PGM3	191.4	99.9	99.7	Immunodeficiency 23, 615816
PHGDH	115.6	100	99.8	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	106.7	98.9	95.3	Muscle glycogenosis, 300559
PHKA2	108.2	100	99.5	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	130.4	99.8	97.5	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	115.2	99.8	97.8	No OMIM phenotype
PHYH	74.6	97.5	90.8	Refsum disease, 266500
PIGA	90.5	90.4	81.3	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	129	99.7	96.4	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGL	121.8	99.9	99.3	CHIME syndrome, 280000
PIGM	165.4	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	111.3	92.6	87.1	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147	100	99.9	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	101.4	91.6	83.8	?Epileptic encephalopathy, early infantile, 55, 617599
PIGQ	125.4	92.6	90.7	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	171.3	98.1	98	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	145.5	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	147.6	100	99.8	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	121.4	100	99.9	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	120.7	99.9	99.1	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000

				Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3R1	129.3	99.7	97.3	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R2	86.2	89.1	86.1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	110.1	100	99.8	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	141.6	99.8	98.4	Corneal fleck dystrophy, 121850
PIP5K1C	107.6	96.3	95.1	Lethal congenital contractural syndrome 3, 611369
PKLR	178.8	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PLA2G5	125.4	100	100	[Fleck retina, familial benign], 228980
PLA2G6	117.5	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	125.6	99.9	97.2	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLCB1	142.8	100	99.7	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	126.1	99.2	95.7	Auriculocondylar syndrome 2, 614669
PLCD1	116.9	99.5	97.1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	155.3	99.5	98.9	Nephrotic syndrome, type 3, 610725
PLCG2	118.9	100	99.8	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLIN1	81	96.2	88.4	Lipodystrophy, familial partial, type 4, 613877
PLOD1	137.9	99.8	97.5	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	108.6	94.7	88.6	Bruck syndrome 2, 609220
PLOD3	100.1	99	96.3	Lysyl hydroxylase 3 deficiency, 612394
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	160.5	99.5	95.6	?Pancreatic lipase deficiency, 614338
PNMT	91.5	99.5	96.6	?Hypertension, essential, 145500
PNP	151.4	100	99.5	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179

PNPLA2	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
PNPLA6	122.1	99.7	98.5	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPO	66.4	100	98.3	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	139.4	99.9	97.5	Dowling-Degos disease 2, 615327
POGLUT1	117.4	98.2	93.8	?Muscular dystrophy, limb-girdle, type 2Z, 617232 Dowling-Degos disease 4, 615696
POLR3A	137.4	100	99.9	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	146.4	99.9	98.5	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	127.6	99.7	97.1	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	259.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	205.1	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	155.7	99.7	98.1	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	111.1	98.9	97.5	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	108.7	99.9	98.4	No OMIM phenotype Cardiomyopathy, dilated (Iuso (2018) Am J Hum Genet 102, 1018)
PPM1K	180	100	100	?Maple syrup urine disease, mild variant, 615135
PPOX	96.1	99.7	98.2	Porphyria variegata, 176200
PPT1	144.5	90	87.3	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	125.6	98.1	91.6	cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200

PRKCSH	135.2	99.7	96.3	Polycystic liver disease 1, 174050
PRODH	83.8	84.9	82.3	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	149.5	100	100	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PSAP	114.4	99.9	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.2	91.4	75.8	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSPH	128.9	98.8	95.4	Phosphoserine phosphatase deficiency, 614023
PTEN	143.2	99.6	96	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome, 0 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTGIS	126.7	96.7	94.6	Hypertension, essential, 145500
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTS	107.2	99.6	94.1	Hyperphenylalaninemia, BH4-deficient, A, 261640
PYCR1	86.3	99.4	94.3	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	158.6	100	100	Glycogen storage disease VI, 232700
PYGM	127.1	100	99.9	McArdle disease, 232600
QDPR	92.3	100	99.5	Hyperphenylalaninemia, BH4-deficient, C, 261630

RBCK1	104.1	99.2	94.9	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	94.4	100	98.4	Leber congenital amaurosis 13, 612712
RDH5	160.2	100	99.7	Fundus albipunctatus, 136880
RFT1	108.3	99.8	97.3	Congenital disorder of glycosylation, type In, 612015
RPE65	130.3	100	99.3	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPIA	113.1	94.3	90.9	?Ribose 5-phosphate isomerase deficiency, 608611
SARDH	120.4	95	92.8	[Sarcosinemia], 268900
SAT1	141.1	100	99.2	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SC5D	198.4	100	99.2	Lathosterolosis, 607330
SCARB2	121.2	100	99.9	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	106.9	99.6	96.5	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	143.2	98.6	96.3	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	161.1	97.5	96.4	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SELENBP1	140.5	100	100	No OMIM phenotype Extraoral halitosis (Pol (2018) Nat Genet 50,120)
SEPSECS	159.3	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	129	95.1	93.6	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SI	98.1	96.2	87.1	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	110	100	98.9	No OMIM phenotype
SLC16A1	157.1	99.9	98.8	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	119.6	96.8	92.9	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC22A12	105.7	100	99.7	Hypouricemia, renal, 220150
SLC22A5	153.3	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A1	71	92.2	87	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	110.7	95.7	92.3	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	192.5	98.8	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970

SLC25A19	88.6	99.9	98.3	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	110.3	100	99.7	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	114	100	98.9	No OMIM phenotype ?Synpolydactyly (Meyertholen (2012) Mol Syndromol 3 25)
SLC25A32	117	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	111.4	99.8	98.1	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	116.9	97.8	94.2	No OMIM phenotype Mitochondrial myopathy (Shamseldin (2016) Hum Genet 135,21)
SLC2A1	190.1	92.9	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	178.4	100	99.9	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	119	99.2	96.2	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	164.4	100	100	Hypermanganesemia with dystonia 1, 613280
SLC33A1	140.9	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC35A1	124	99.9	97.7	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	108.8	99.7	96.8	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	50.2	78.1	71.9	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	230.2	99.9	98.4	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	115.4	95.7	90.4	Schneckenbecken dysplasia, 269250
SLC37A4	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A14	107.7	99.8	97.9	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	81.8	99.2	96	Acrodermatitis enteropathica, 201100
SLC39A8	128.5	100	99.7	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	162.8	100	99.5	Cystinuria, 220100
SLC46A1	106	99.4	96.4	Folate malabsorption, hereditary, 229050

SLC52A1	219.3	100	100	Riboflavin deficiency, 615026
SLC52A2	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.6	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	140	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	118.7	100	100	Renal glucosuria, 233100
SLC6A19	149.1	100	99.3	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A8	56.5	89.8	79.1	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	123.9	100	99.9	Lysinuric protein intolerance, 222700
SLC7A9	125.5	99.9	99	Cystinuria, 220100
SLCO1B1	46	92.8	84.4	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	48.9	94.9	79.5	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	123.5	99.6	97.9	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	67.8	88.3	73.9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SOD1	161.9	100	100	Amyotrophic lateral sclerosis 1, 105400
SPR	166.5	98.9	90	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	115.5	99	93.9	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	160.2	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SRD5A2	77.6	100	96.4	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	135.9	100	99.7	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SSR4	89.8	100	98.8	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	144.5	100	99.9	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	121.9	84.4	84.2	Salt and pepper developmental regression syndrome, 609056
STAR	124	100	100	Lipoid adrenal hyperplasia, 201710
STS	91.6	99.7	97.8	Ichthyosis, X-linked, 308100
STT3A	156.2	100	100	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	125.1	99.2	96	?Congenital disorder of glycosylation, type Ix, 615597
SUCLA2	64.9	93.3	82.8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

SUCLG1	101.3	99.6	95.4	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	57.8	91.1	78.7	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SUGCT	129.1	94.1	87.6	Glutaric aciduria III, 231690
SUMF1	103.3	98.6	91.1	Multiple sulfatase deficiency, 272200
SUOX	212.6	100	100	Sulfite oxidase deficiency, 272300
TALDO1	130.5	100	99.9	Transaldolase deficiency, 606003
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	143.1	100	100	Tyrosinemia, type II, 276600
TAZ	94	99.9	98.8	Barth syndrome, 302060
TBXAS1	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TCN2	175.6	100	100	Transcobalamin II deficiency, 275350
TECR	94.6	99.9	97.9	Mental retardation, autosomal recessive 14, 614020
TH	68.2	97.6	88.7	Segawa syndrome, recessive, 605407
TIMM50	108.2	98.8	95.3	3-methylglutaconic aciduria, type IX, 617698
TK2	105.7	93.4	89.4	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	114.1	98.7	97.7	Short stature, developmental delay, and congenital heart defects, 617044
TMEM165	113.9	99.8	98.1	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	105.1	100	99.9	Congenital disorder of glycosylation, type IIp, 616829
TMEM5	120.5	96.8	92.9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM70	138.7	94.6	90.3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	100.9	99.9	97.6	{Autism, susceptibility to, X-linked 6}, 300872
TPI1	103	99.2	96.7	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPMT	45.5	94.7	78.6	{Thiopurines, poor metabolism of, 1}, 610460
TPP1	146.3	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAK1	152.3	99	98.1	No OMIM phenotype
TRAPPC11	126.2	99.4	96.4	Muscular dystrophy, limb-girdle, type 2S, 615356

TREH	143.1	98	93.5	Trehalase deficiency, 612119
TUSC3	136.4	100	98.3	Mental retardation, autosomal recessive 7, 611093
TYMP	95.2	98.3	85	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	185.3	100	100	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYRP1	176.9	100	99.9	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UGT1A1	240.6	100	100	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMPS	173.6	99.3	97.2	Orotic aciduria, 258900
UPB1	157.4	100	100	Beta-ureidopropionase deficiency, 613161
UROC1	132	99.9	99	?Urocanase deficiency, 276880
UROD	163.1	99.8	97.9	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	108.3	100	99.9	Porphyria, congenital erythropoietic, 263700
VPS13B	143.8	98.6	96.8	Cohen syndrome, 216550
XDH	109.1	100	99.9	Xanthinuria, type I, 278300
XYLT1	132.5	90.4	87.1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.3	98.9	94.9	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 : September 11th, 2018.

This list is accurate for panel version DG 2.14

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