METABOLIC DISORDERS GENE PANEL DG 3.1.0 (703 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AASS	100	99,7	100	100	Hyperlysinemia, 238700
ABAT	100	99,4	100	100	GABA-transaminase deficiency, 613163
ABCC8	100	99,8	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD1	75,8	71,6	100	100	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD2	100	99,8	100	100	No OMIM disease ID
ABCD3	99,8	97,7	100	100	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	99,9	98,6	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100	100	100	100	Sitosterolemia 2, 618666
ABCG8	99,1	97,3	100	100	{Gallbladder disease 4}, 611465 Sitosterolemia 1, 210250
ABHD12	91,2	85,2	100	99,4	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100	100	100	100	Chanarin-Dorfman syndrome, 275630
ACACA	98,4	98,1	100	100	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100	100	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	99,8	99	100	100	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	99,9	98,2	100	100	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100	99,2	100	100	2-methylbutyrylglycinuria, 610006
ACADVL	99,4	97,3	100	100	VLCAD deficiency, 201475
ACAT1	99,9	97,6	100	100	Alpha-methylacetoacetic aciduria, 203750

ACAT2	100	100	100	100	?ACAT2 deficiency, 614055
ACBD5	100	99,2	100	100	Retinal dystrophy with leukodystrophy, 618863
ACO2	96,3	90,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100	99,9	100	100	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100	99,2	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACSF3	100	99,9	100	100	Combined malonic and methylmalonic aciduria, 614265
ACSL4	98,7	94,6	100	100	Mental retardation, X-linked 63, 300387
ACY1	100	98,8	100	100	Aminoacylase 1 deficiency, 609924
ADA	100	99,7	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	100	99,9	100	100	No OMIM disease ID
ADCY5	95,1	91,2	99,2	98	Dyskinesia, familial, with facial myokymia, 606703
ADK	84,1	81	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	99,2	98,7	100	100	Adenylosuccinase deficiency, 103050
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
AGK	90,6	88,6	91,2	91,2	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	100	99,4	100	100	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	99,6	96,1	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	99,3	95,4	100	99,9	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHCY	100	99,2	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	100	100	100	100	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	98,9	94,9	100	99,9	Reticular dysgenesis, 267500
AKR1C1	95,9	89,7	100	100	No OMIM disease ID
AKR1D1	100	99,4	100	100	Bile acid synthesis defect, congenital, 2, 235555
ALAD	99,3	94,1	100	100	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740

ALAS2	98,9	94,9	100	100	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ALDH18A1	100	99,9	100	100	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	97,2	94,5	100	99,9	Microphthalmia, isolated 8, 615113
ALDH2	100	100	100	100	Alcohol sensitivity, acute, 610251 {Hangover, susceptibility to}, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Sublingual nitroglycerin, susceptibility to poor response to}, 0
ALDH3A2	88,8	88,1	93,2	93,2	Sjogren-Larsson syndrome, 270200
ALDH4A1	100	99,4	100	100	Hyperprolinemia, type II, 239510
ALDH5A1	91	81,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100	99,9	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	94,4	88,8	100	100	Epilepsy, pyridoxine-dependent, 266100
ALDOA	98,9	96,9	100	100	Glycogen storage disease XII, 611881
ALDOB	99,4	96,6	100	100	Fructose intolerance, hereditary, 229600
ALG1	53	45,8	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100	100	100	100	No OMIM disease ID
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100	100	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	98,4	92,6	100	99,6	Developmental and epileptic encephalopathy 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG14	100	99,9	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031
ALG2	100	100	100	100	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,6	94,8	100	100	Congenital disorder of glycosylation, type Ic, 603147
ALG8	97,2	95,6	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874

ALG9	100	99,7	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALOX12B	100	100	100	100	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	100	100	100	100	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
AMACR	100	100	100	100	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMN	89,7	80	100	100	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	99,9	98,6	100	100	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	99,9	98,5	100	100	[AMP deaminase deficiency, erythrocytic], 612874
AMT	100	100	100	100	Glycine encephalopathy, 605899
AP1S1	99,9	99,5	100	100	MEDNIK syndrome, 609313
AP3B2	93,3	89,5	99,8	98,6	Developmental and epileptic encephalopathy 48, 617276
APOA5	100	99,9	100	99,5	{Hypertriglyceridemia, susceptibility to}, 145750 Hyperchylomicronemia, late-onset, 144650
APOC2	100	100	100	100	Hyperlipoproteinemia, type Ib, 207750
APRT	100	99,5	100	100	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	92,9	92,9	92,9	92,9	Argininemia, 207800
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ARSB	97	88,7	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	99,7	98,6	100	100	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	100	99,6	100	100	Argininosuccinic aciduria, 207900
ASNS	99,4	95,2	100	100	Asparagine synthetase deficiency, 615574
ASPA	99,9	98,3	100	100	Canavan disease, 271900
ASS1	95,4	87,9	100	100	Citrullinemia, 215700
ATIC	99,9	99,3	100	100	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP6AP1	98,2	92,1	100	100	Immunodeficiency 47, 300972

ATP6AP2	94,1	76,6	100	100	Congenital disorder of glycosylation, type IIr, 301045 Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP6V0A2	100	99,5	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	99,9	98,7	100	100	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1E1	93,1	88,3	100	100	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	99	96,9	100	100	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8B1	96,5	94	100	100	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
AUH	100	99,8	100	100	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	100	99,8	100	100	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	93,8	89,4	92,5	92,5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	75,7	69,7	89,8	81,6	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	99,9	98,2	94,8	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	99,6	96,3	99,9	99,2	Peters-plus syndrome, 261540
B4GALT1	100	99,8	100	100	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	99,8	97,4	99,9	98,6	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	99,8	98,4	100	100	Hypercholanemia, familial, 607748
BCAT1	100	100	100	100	No OMIM disease ID
BCAT2	100	100	100	100	?Hypervalinemia or hyperleucine-isoleucinemia, 618850
BCKDHA	99,9	99,2	100	100	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,5	94,4	100	100	Maple syrup urine disease, type Ib, 248600
BCKDK	100	100	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923

BCO1	100	100	100	100	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	100	99,4	100	100	Hyperbiliverdinemia, 614156
BMP2	100	100	100	100	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877
BPGM	100	100	100	100	Erythrocytosis, familial, 8, 222800
BTD	83,1	83	83,1	83,1	Biotinidase deficiency, 253260
C1GALT1C1	100	99,5	100	100	Tn polyagglutination syndrome, somatic, 300622
CA5A	87,4	85,2	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	100	99,2	100	100	Developmental and epileptic encephalopathy 50, 616457
CANT1	100	99,9	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAT	100	100	100	100	Acatalasemia, 614097
CBLIF	100	99,7	100	100	Intrinsic factor deficiency, 261000
CBS	99,8	98,3	100	100	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCDC115	95,3	90	100	100	Congenital disorder of glycosylation, type IIo, 616828
CD320	100	99,8	100	100	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CEL	89,7	88	100	99,8	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	99,5	96,9	100	100	Retinitis pigmentosa 26, 608380
CERS3	99,9	98,9	100	100	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	99,6	97,9	100	100	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 {Pancreatitis, hereditary}, 167800 Sweat chloride elevation without CF, 0 {Hypertrypsinemia, neonatal}, 0
CHIT1	99,7	98,1	100	100	[Chitotriosidase deficiency], 614122
CHKB	100	99,7	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,4	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100	100	100	100	Macular corneal dystrophy, 217800

CHSY1	97,2	95,7	99,7	98	Temtamy preaxial brachydactyly syndrome, 605282
CLCN7	99,7	98,4	100	100	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLN3	92,5	91,8	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69,3	66,3	72,1	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,9	97,1	100	100	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPB	94,9	94,9	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CMAS	99,9	97,8	100	100	No OMIM disease ID
COG1	100	100	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG2	99,9	98,5	100	100	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100	99,9	100	100	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	99,7	97,6	100	100	Congenital disorder of glycosylation, type IIi, 613612
COG6	99,1	93,9	100	100	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	100	100	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	99,9	98,6	100	100	Congenital disorder of glycosylation, type IIh, 611182
COMT	100	99,9	100	100	{Schizophrenia, susceptibility to}, 181500 {Panic disorder, susceptibility to}, 167870
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,9	89,3	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100	100	100	100	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	99,9	98,4	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,8	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100	99,5	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100	99,3	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	97,9	100	100	Coenzyme Q10 deficiency, primary, 5, 614654

CP	94,8	88,9	100	100	[Hypoceruloplasminemia, hereditary], 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CPOX	99,9	95,4	100	100	Harderoporphyria, 618892 Coproporphyria, 121300
CPS1	100	99,9	100	100	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPT1A	100	98,9	100	100	CPT deficiency, hepatic, type IA, 255120
CPT2	98,2	97,8	100	100	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRAT	100	99,8	100	100	?Neurodegeneration with brain iron accumulation 8, 617917
CRPPA	98,5	94,8	100	99,4	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CTH	100	100	100	100	Cystathioninuria, 219500
CTNS	100	99,8	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTSA	100	100	100	100	Galactosialidosis, 256540
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	100	99,9	100	100	Pycnodysostosis, 265800
CUBN	99,7	98,3	100	100	[Proteinuria, chronic benign], 618884 Imerslund-Grasbeck syndrome 1, 261100
CYB561	92,8	92,6	100	99,9	Orthostatic hypotension 2, 618182
CYB5R3	98,4	98	99,8	98,9	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	99,3	96,1	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	100	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900

CYP11B2	100	100	100	100	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Aldosterone to renin ratio raised, 0 {Low renin hypertension, susceptibility to}, 0
CYP17A1	100	99,5	100	100	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	98,8	96,8	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	100	100	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	97,8	88,4	100	100	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP27A1	98,9	96,7	100	100	Cerebrotendinous xanthomatosis, 213700
CYP27B1	99,9	99,3	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2R1	99,4	95,6	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	94,8	91,5	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	98	92,8	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99,2	97,2	100	100	D-2-hydroxyglutaric aciduria, 600721
DAO	100	100	100	100	No OMIM disease ID
DBH	100	100	100	100	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	99,8	98	100	100	Maple syrup urine disease, type II, 248600
DCXR	98,6	93,6	100	100	[Pentosuria], 260800
DDC	99,7	96,4	100	100	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	97,9	95,8	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	100	99,9	100	100	?Congenital disorder of glycosylation, type Ir, 614507
DEGS1	100	100	100	100	Leukodystrophy, hypomyelinating, 18, 618404
DGAT1	91,9	87,6	99,7	98,6	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	99,8	98,1	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008
DGUOK	100	99,4	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880

DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	99	95	95,2	95,2	Retinitis pigmentosa 59, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861
DHFR	92,1	78,9	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	100	100	100	100	Miller syndrome, 263750
DLD	100	99,7	100	100	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	100	99,7	100	100	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	87,4	87,4	100	100	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	98,9	96,2	100	100	3-methylglutaconic aciduria, type V, 610198
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	98,1	94,5	100	100	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	99,2	99	99,7	99,2	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	100	100	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	98,2	91,3	99,7	97,1	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	98,7	100	100	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992
DPYD	99,7	97,7	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100	99,9	100	100	Dihydropyrimidinuria, 222748
DTYMK	100	99,8	100	100	No OMIM disease ID
EBP	99,7	95,8	100	100	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960

ECHS1	99,9	99	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL1	99,8	97,6	100	100	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100	99,5	100	100	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	100	99,9	100	100	?Glycogen storage disease XIII, 612932
EOGT	79,4	78,4	91,9	89	Adams-Oliver syndrome 4, 615297
EPHX1	99,9	98,8	100	100	?Hypercholanemia, familial, 607748
EPHX2	99,5	96,2	100	100	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
ETFA	100	100	100	100	Glutaric acidemia IIA, 231680
ETFB	100	99,8	100	100	Glutaric acidemia IIB, 231680
ETFDH	100	99,8	100	100	Glutaric acidemia IIC, 231680
ETHE1	99,9	97,4	100	100	Ethylmalonic encephalopathy, 602473
EXT1	99,9	98,4	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100	99,3	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EYA1	99,9	99,7	100	100	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
FA2H	92	83,1	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FAH	100	100	100	100	Tyrosinemia, type I, 276700
FAR1	97,6	92,8	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBP1	93,7	93,3	93,7	93,7	Fructose-1,6-bisphosphatase deficiency, 229700
FCSK	97,7	95,4	100	100	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	97,7	96	100	100	Squalene synthase deficiency, 618156
FECH	100	100	100	100	Protoporphyria, erythropoietic, 1, 177000
FH	92,1	88,3	100	100	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKRP	100	100	100	99,9	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	99,7	97	100	100	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	100	99,8	100	100	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	99,9	99,7	100	100	Trimethylaminuria, 602079
FOLR1	100	100	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	95,7	91	100	100	Glutamate formiminotransferase deficiency, 229100
FUCA1	100	99,9	100	100	Fucosidosis, 230000
FUT2	100	100	100	100	{Vitamin B12 plasma level QTL1}, 612542 [Bombay phenotype, digenic], 616754 {Norwalk virus infection, resistance to}, 0
FUT6	100	100	100	100	[Fucosyltransferase 6 deficiency], 613852
FUT8	100	99,2	100	100	Congenital disorder of glycosylation with defective fucosylation 1, 618005
G6PC	100	100	100	100	Glycogen storage disease Ia, 232200
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	99,3	98	100	100	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	100	99,9	100	100	Glycogen storage disease II, 232300
GAD1	100	99,9	100	100	?Cerebral palsy, spastic quadriplegic, 1, 603513 Developmental and epileptic encephalopathy 89, 619124
GALC	99,8	98,3	100	100	Krabbe disease, 245200
GALE	100	100	100	100	Galactose epimerase deficiency, 230350
GALK1	100	99,1	100	100	Galactokinase deficiency with cataracts, 230200
GALM	100	99,9	100	100	Galactosemia IV, 618881
GALNS	100	99,8	100	100	Mucopolysaccharidosis IVA, 253000
GALNT2	99,6	97	100	100	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	99,8	99	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100	99,7	100	100	Galactosemia, 230400
GAMT	93,1	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736

GANAB	99,9	99	100	100	Polycystic kidney disease 3, 600666
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GBA2	100	99,7	100	100	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GCDH	100	99,2	100	100	Glutaricaciduria, type I, 231670
GCH1	99,9	95,5	100	100	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCK	95,4	95,4	92,5	92,2	Diabetes mellitus, noninsulin-dependent, late onset, 125853 MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCLC	99,8	98	100	100	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	99,6	95,8	100	100	{Myocardial infarction, susceptibility to}, 608446
GCSH	75,7	68,9	100	100	?Glycine encephalopathy, 605899
GFPT1	100	99,4	100	100	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGPS1	100	100	100	100	No OMIM disease ID
GK	88,9	70,4	100	99,9	Glycerol kinase deficiency, 307030
GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	99,9	97,4	100	100	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	89,9	82	100	99,9	Glycine encephalopathy, 605899
GLRA1	100	99,8	100	100	Hyperekplexia 1, 149400

GLRX5	97,3	89,1	99,6	95,4	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	96,3	87,2	100	99,9	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Global developmental delay, progressive ataxia, and elevated glutamine, 618412 Developmental and epileptic encephalopathy 71, 618328
GLUD1	94,2	82,9	100	100	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	74,4	73,2	100	100	Glutamine deficiency, congenital, 610015
GLYCTK	98,8	97,3	100	100	D-glyceric aciduria, 220120
GM2A	100	100	100	100	GM2-gangliosidosis, AB variant, 272750
GMPPA	100	100	100	100	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100	100	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	99,1	96,1	100	100	No OMIM disease ID
GNE	100	99,7	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100	100	100	100	Glycine N-methyltransferase deficiency, 606664
GNPAT	99,7	97,3	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,1	94,3	100	99,9	Mucolipidosis III gamma, 252605
GNS	98,4	94,8	100	100	Mucopolysaccharidosis type IIID, 252940
GOT1	100	99,3	100	100	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	97,5	90,9	100	100	Developmental and epileptic encephalopathy 82, 618721
GPD1	100	100	100	100	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100	99,8	100	100	Brugada syndrome 2, 611777
GPHN	100	99,5	100	100	Molybdenum cofactor deficiency C, 615501
GPI	100	100	100	100	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100	100	100	100	Hyperlipoproteinemia, type 1D, 615947
GPT2	99,2	93,6	100	99,8	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	95,9	86,4	100	100	Hemolytic anemia due to glutathione peroxidase deficiency, 614164

GRHPR	84,2	81,3	100	99,3	Hyperoxaluria, primary, type II, 260000
GSS	96,5	96,4	100	100	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GUSB	92,9	91,7	100	100	Mucopolysaccharidosis VII, 253220
GYG1	99,9	99,2	100	100	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100	98,6	100	100	Glycogen storage disease 0, muscle, 611556
GYS2	99,8	99	100	100	Glycogen storage disease 0, liver, 240600
H6PD	99	99	100	100	Cortisone reductase deficiency 1, 604931
HADH	99	97,5	100	100	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HADHB	98,8	89,7	100	100	Trifunctional protein deficiency, 609015
HAGH	100	99,7	98,7	96,1	[Glyoxalase II deficiency], 614033
HEXA	93,8	93,3	100	100	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	99,6	96,9	100	99,9	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	100	99,7	100	100	{Porphyria variegata, susceptibility to}, 176200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200
HGD	100	100	100	100	Alkaptonuria, 203500
HGSNAT	86,4	86,3	91,2	89,3	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	94,4	91,2	100	100	No OMIM disease ID
HIBCH	98,2	88,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	100	100	100	100	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	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HMBS	99,9	99,4	100	100	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	100	99,8	100	100	HMG-CoA lyase deficiency, 246450
HMGCS2	100	99,6	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	98,4	89,9	100	100	{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 Heme oxygenase-1 deficiency, 614034
HNF1A	100	99,8	100	100	{Diabetes mellitus, insulin-dependent}, 222100 MODY, type III, 600496 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HNF4A	99,9	99	100	100	{Diabetes mellitus, noninsulin-dependent}, 125853 MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HOGA1	100	96,4	100	100	Hyperoxaluria, primary, type III, 613616
HPD	100	100	100	100	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPDL	100	100	100	100	Spastic paraplegia 83, autosomal recessive, 619027 Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026
HPRT1	99,3	91,8	100	99,3	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	92,9	84,5	100	100	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD11B1	100	99,6	100	100	Cortisone reductase deficiency 2, 614662
HSD11B2	86	82,7	99,9	98,1	Apparent mineralocorticoid excess, 218030
HSD17B10	100	99,1	100	100	HSD10 mitochondrial disease, 300438
HSD17B3	97,8	97,8	97,8	97,8	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100	99,7	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	99,1	95,5	100	100	Bile acid synthesis defect, congenital, 1, 607765

HTRA2	100	99,9	100	100	{Parkinson disease 13}, 610297 3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100	100	100	100	?Mucopolysaccharidosis type IX, 601492
IDH2	99,7	97,4	100	99,8	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	95,4	95,4	100	100	Retinitis pigmentosa 46, 612572
IDI1	99,8	97,6	100	100	No OMIM disease ID
IDS	99,9	98	100	100	Mucopolysaccharidosis II, 309900
IDUA	93,7	86,8	100	100	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IMPAD1	100	100	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	87,9	80,2	100	100	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
INPP5E	97,1	92,7	100	100	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPPL1	98,4	94,5	99,9	99,7	Opsismodysplasia, 258480
INSR	97,8	94,7	99,9	99,2	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
IREB2	100	99,8	100	100	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
ITCH	91,6	91,3	95,9	95	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850
IVD	100	100	100	100	Isovaleric acidemia, 243500
KCNA2	100	99,6	100	100	Developmental and epileptic encephalopathy 32, 616366
KCNJ11	100	100	100	100	Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KMT2A	100	99,9	99,9	99,4	Wiedemann-Steiner syndrome, 605130
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
L2HGDH	99	97,2	100	100	L-2-hydroxyglutaric aciduria, 236792

LAMP2	99,2	95,6	100	100	Danon disease, 300257
LARGE1	100	99,6	100	100	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	99	93,8	100	100	Norum disease, 245900 Fish-eye disease, 136120
LCT	99,8	98,5	100	100	Lactase deficiency, congenital, 223000
LDHA	95	91,7	100	100	Glycogen storage disease XI, 612933
LDHB	94,7	84,3	100	100	[Lactate dehydrogenase-B deficiency], 614128
LFNG	87,9	86,4	92,2	87,7	Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	100	99,1	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	99,2	95,2	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100	99,4	100	100	[High density lipoprotein cholesterol level QTL 12], 612797 Hepatic lipase deficiency, 614025 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPT1	100	99,9	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	94,9	75,2	100	100	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	94,7	90,2	96,1	96,1	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100	99,6	100	100	Lipase deficiency, combined, 246650
LPIN1	99,6	97,3	100	100	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	100	100	100	100	Majeed syndrome, 609628
LPL	100	100	100	100	Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 238600 Combined hyperlipidemia, familial, 144250
LRAT	100	100	100	100	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	74,2	68,5	100	100	Leukotriene C4 synthase deficiency, 614037
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
MAN1B1	100	99,7	100	99,9	Rafiq syndrome, 614202
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	99,9	99,1	100	100	No OMIM disease ID
MANBA	87,8	86,5	100	100	Mannosidosis, beta, 248510

MAOA	100	99,7	99,8	98,5	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAT1A	99,7	97,7	100	100	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MBOAT7	100	99,5	100	100	Mental retardation, autosomal recessive 57, 617188
MCCC1	100	99,8	100	100	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,9	98,4	100	100	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100	100	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	99,8	98,4	100	100	Mucolipidosis IV, 252650
MDH1	100	99	100	100	?Developmental and epileptic encephalopathy 88, 618959
MFSD2A	99,7	98,5	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100	99,7	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100	100	100	100	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	100	99,5	100	100	{Thyroid carcinoma, follicular}, 188470
MLYCD	96	90,4	100	98,9	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100	100	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100	99,6	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94,4	83,5	89,7	89,7	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MMUT	99,8	98,3	100	100	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	99,8	97,7	100	100	Xanthinuria, type II, 603592
MOCS1	99,2	95,1	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,6	99,5	100	100	Molybdenum cofactor deficiency B, 252160
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	100	100	100	100	Congenital disorder of glycosylation, type If, 609180
MPI	100	99,9	100	100	Congenital disorder of glycosylation, type Ib, 602579
MRPL44	99,9	98,7	100	100	?Combined oxidative phosphorylation deficiency 16, 615395

MRPS36	95,2	77,6	100	100	No OMIM disease ID
MSMO1	96,3	88,9	100	100	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	100	99,5	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	97,3	96	100	100	{Schizophrenia, susceptibility to}, 181500 Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTM1	99	93,3	100	100	Myotubular myopathy, X-linked, 310400
MTMR2	100	99	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	100	100	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100	99,6	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cbl E type, 236270
MVK	90,9	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
NADK2	99,9	97,2	99	96,3	2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	100	100	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	92,9	89,9	99,9	99,2	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	99,7	95	100	100	N-acetylglutamate synthase deficiency, 237310
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NAXD	100	100	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100	99,8	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NGLY1	100	99,8	100	100	Congenital disorder of deglycosylation, 615273
NMNAT1	100	99,2	98,3	95,6	Leber congenital amaurosis 9, 608553

NNT	96,4	95,9	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NPC1	99,6	98,7	100	100	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100	99,6	100	100	Niemann-pick disease, type C2, 607625
NPL	100	99,9	100	100	No OMIM disease ID
NSD1	100	99,9	100	100	Sotos syndrome 1, 117550
NSDHL	100	98,7	100	100	CHILD syndrome, 308050 CK syndrome, 300831
NT5C3A	97,8	88,2	100	100	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100	99,9	100	100	Calcification of joints and arteries, 211800
NUS1	60	44,5	100	100	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	85,2	76,3	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	99,9	98,6	100	99,9	Lowe syndrome, 309000 Dent disease 2, 300555
OGDH	100	99,9	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA3	100	99	100	100	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPLAH	100	99,8	100	100	5-oxoprolinase deficiency, 260005
OTC	100	100	100	100	Ornithine transcarbamylase deficiency, 311250
OXCT1	99,8	98,1	100	100	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	100	100	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PANK2	100	99,3	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	99,8	97,3	100	100	Pyruvate carboxylase deficiency, 266150
PCBD1	100	99,6	100	99,7	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,5	96,7	100	100	Propionicacidemia, 606054
PCCB	97,9	96	98,7	96,2	Propionicacidemia, 606054
PCK1	100	100	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100	100	100	100	PEPCK deficiency, mitochondrial, 261650
PCYT1A	98,9	95,5	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940

PCYT2	99,8	97,1	100	98,8	Spastic paraplegia 82, autosomal recessive, 618770
PDSS1	94,7	87,6	97,3	96,6	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,8	97,1	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	100	98,8	100	100	Prolidase deficiency, 170100
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	100	99,6	100	100	Peroxisome biogenesis disorder 14B, 614920
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	96,7	90,8	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,9	98,5	100	100	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100	100	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100	100	100	100	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100	99,3	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	100	99,5	100	100	Glycogen storage disease VII, 232800
PGAM2	100	100	100	100	Glycogen storage disease X, 261670
PGAP1	99	94,4	100	100	Mental retardation, autosomal recessive 42, 615802

PGAP2	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	63,5	59,6	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	92,8	79,3	100	100	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94,2	94,2	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PGM3	100	99,8	91,7	91,7	Immunodeficiency 23, 615816
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	99,2	95,3	100	99,9	Muscle glycogenosis, 300559
PHKA2	100	99,7	100	99,6	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	99,9	99,2	100	100	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	99,9	97,8	100	100	No OMIM disease ID
PHKG2	100	99,9	100	100	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency, 0
PHYH	100	99,6	100	100	Refsum disease, 266500
PI4K2A	91,9	86,4	100	100	No OMIM disease ID
PIGA	93,8	86,7	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	99,9	97,8	100	100	Developmental and epileptic encephalopathy 80, 618580
PIGC	99,2	90,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGL	100	100	100	100	CHIME syndrome, 280000
PIGM	100	100	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	93,8	91,5	98,8	98,8	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,8	87,3	100	100	Developmental and epileptic encephalopathy 55, 617599
PIGQ	92,8	90,8	100	100	Developmental and epileptic encephalopathy 77, 618548
PIGT	98,1	98,1	100	100	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100	99,8	100	100	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 6, 616809

PIK3CA	98	97,8	100	100	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	99,8	99	100	100	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	90,7	89,6	99,3	96,1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100	99,9	100	100	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	99,9	99,4	100	100	Corneal fleck dystrophy, 121850
PIP5K1C	98	95,8	99,9	99,8	Lethal congenital contractural syndrome 3, 611369
PKLR	100	99,2	100	100	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PLA2G5	100	100	100	100	[Fleck retina, familial benign], 228980
PLA2G6	92,2	90,7	92,3	92,3	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLA2G7	99,9	99	100	100	{Asthma, susceptibility to}, 600807 Platelet-activating factor acetylhydrolase deficiency, 614278 {Atopy, susceptibility to}, 147050
PLCB1	100	99,8	100	100	Developmental and epileptic encephalopathy 12, 613722
PLCB4	99,9	98,8	100	100	Auriculocondylar syndrome 2, 614669
PLCD1	99,9	97,8	100	100	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	99,9	99,3	100	100	Nephrotic syndrome, type 3, 610725
PLCG2	100	99,8	100	100	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878

PLIN1	99,6	94,9	100	99,5	Lipodystrophy, familial partial, type 4, 613877
PLOD1	100	98,4	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,3	97,3	100	100	Bruck syndrome 2, 609220
PLOD3	99,8	98	100	100	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	98,2	90,1	100	100	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	100	99,8	100	100	?Pancreatic lipase deficiency, 614338
PNMT	99,6	96,7	100	100	No OMIM disease ID
PNP	99,8	98,9	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	99,7	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
PNPO	99,9	97,7	100	100	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	100	99	100	100	Dowling-Degos disease 2, 615327
POGLUT1	99,4	94,6	100	100	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLR3A	100	99,7	100	100	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,9	98,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	100	99,9	100	100	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	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100	100	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	99,3	97,5	100	100	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	99,4	96,4	100	100	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	99,8	99,5	100	100	Cardiomyopathy, dilated, 2C, 618189
PPM1K	100	100	100	100	?Maple syrup urine disease, mild variant, 615135
PPOX	99,7	96,8	100	100	Porphyria variegata, 176200
PPT1	90,3	90,3	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	99,1	96,7	100	99,4	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKCSH	99,8	95,4	100	100	Polycystic liver disease 1, 174050
PRODH	85	80,6	100	100	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	86,4	86,4	100	100	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	100	100	100	100	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	95,3	81,6	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSPH	100	100	100	100	Phosphoserine phosphatase deficiency, 614023
PTEN	99,5	97	100	100	Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 {Meningioma}, 607174
PTGIS	98,2	95,1	100	100	Hypertension, essential, 145500
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785

PTS	99,9	99,1	100	100	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS3	100	100	100	100	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PYCR1	99,9	97,7	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,1	100	100	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100	100	100	100	Glycogen storage disease VI, 232700
PYGM	100	99,9	100	100	McArdle disease, 232600
QDPR	100	99,7	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
RBCK1	99,9	98,2	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	100	98,6	100	100	Leber congenital amaurosis 13, 612712
RDH5	100	99,9	100	100	Fundus albipunctatus, 136880
RFT1	99,8	99,6	100	100	Congenital disorder of glycosylation, type In, 612015
RINT1	99,9	98,6	100	100	Infantile liver failure syndrome 3, 618641
RPE65	99,8	97,8	100	100	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
RPIA	98,6	94,9	100	100	Ribose 5-phosphate isomerase deficiency, 608611
RPN2	100	100	100	100	No OMIM disease ID
RXYLT1	99,5	96,8	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SARDH	93,7	91,7	91,4	91,4	[Sarcosinemia], 268900
SAT1	99,9	98,5	100	99,9	No OMIM disease ID
SC5D	100	99,5	100	100	Lathosterolosis, 607330
SCARB2	100	99,8	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	100	99,2	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	99,9	99,3	100	100	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SELENBP1	100	100	100	100	Extraoral halitosis due to MTO deficiency, 618148
SEPSECS	100	100	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,9	99,5	100	100	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	94,4	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900

SI	99,2	96,1	100	100	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	99,7	98	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC12A1	96,2	96,1	96,2	96,2	Bartter syndrome, type 1, 601678
SLC13A3	99,4	97,5	100	100	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC16A1	100	99,3	100	100	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC17A5	99,6	97	100	100	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC18A2	100	99,7	100	100	?Parkinsonism-dystonia, infantile, 2, 618049
SLC22A12	100	99,8	100	100	Hypouricemia, renal, 220150
SLC22A5	100	100	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A1	95,8	88,6	99,5	97,8	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A13	100	99,7	100	100	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	99,8	98,1	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100	98,5	100	100	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A20	100	100	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100	99,7	100	100	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A32	100	100	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	97,9	95,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	96,5	93,2	100	100	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC28A1	100	98,8	100	100	[Uridine-cytidineuria], 618477
SLC2A1	92,8	92,8	100	100	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 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A2	100	100	100	100	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853

SLC2A9	99,8	96,1	100	100	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC30A10	100	100	100	100	Hypermanganesemia with dystonia 1, 613280
SLC33A1	99,9	98,9	100	100	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	100	99,7	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	99,9	98,4	100	100	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	80,7	78,6	81,1	81	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	99,9	98,7	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100	97,7	100	100	Schneckenbecken dysplasia, 269250
SLC37A4	100	99,2	100	100	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC39A14	100	99,4	93,5	93,5	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	99,5	95,5	100	100	Acrodermatitis enteropathica, 201100
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100	99,8	96,6	96,6	Cystinuria, 220100
SLC44A1	98,2	98,2	100	100	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC52A1	100	100	100	100	Riboflavin deficiency, 615026
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC5A1	100	100	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	100	100	100	100	Renal glucosuria, 233100
SLC6A19	100	100	100	100	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A5	100	100	100	100	Hyperekplexia 3, 614618
SLC6A8	93,5	81,6	100	99,8	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100	100	100	100	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700

SLC7A9	100	99,9	100	100	Cystinuria, 220100
SLCO1B1	99,2	93,7	100	100	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	98,8	90,8	100	100	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	100	100	100	100	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	91,5	78,5	100	99,9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNX14	99,6	95,9	100	100	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOD1	100	99,9	100	100	Amyotrophic lateral sclerosis 1, 105400 Spastic tetraplegia and axial hypotonia, progressive, 618598
SOD2	100	100	100	100	{Microvascular complications of diabetes 6}, 612634
SPR	99,8	96,3	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	99,2	95,4	100	100	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQOR	100	97,8	100	100	Sulfide:quinone oxidoreductase deficiency, 619221
SRD5A2	99,9	99	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	99,9	99,1	100	100	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SSR4	100	99,7	100	100	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	68,8	68,6	95,3	95,2	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	85	84,2	98,7	98,4	Salt and pepper developmental regression syndrome, 609056
STAR	100	100	100	100	Lipoid adrenal hyperplasia, 201710
STS	97,1	95,5	97,4	97,3	Ichthyosis, X-linked, 308100
STT3A	100	100	100	100	Congenital disorder of glycosylation, type Iw, 615596
STT3B	100	99,6	100	100	?Congenital disorder of glycosylation, type Ix, 615597
STX5	95,8	92,6	100	100	No OMIM disease ID
SUCLA2	89,5	82,2	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	99,9	99,8	100	100	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	96,7	86,3	100	100	No OMIM disease ID
SUGCT	99,9	98,5	100	100	Glutaric aciduria III, 231690

SUMF1	97,5	90,8	100	100	Multiple sulfatase deficiency, 272200
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
TALDO1	100	97,9	100	100	Transaldolase deficiency, 606003
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	100	100	100	100	Tyrosinemia, type II, 276600
TAZ	99,1	95,5	100	100	Barth syndrome, 302060
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	97,6	90,1	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TECR	100	99	100	100	Mental retardation, autosomal recessive 14, 614020
TH	99,3	96,1	100	100	Segawa syndrome, recessive, 605407
TIMM50	98,3	94,4	100	100	3-methylglutaconic aciduria, type IX, 617698
TK2	99,2	96,3	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100	99,5	100	100	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98,7	97,8	98,7	98,7	Short stature, developmental delay, and congenital heart defects, 617044
TMEM106B	99,9	98,8	100	100	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	100	99,9	100	100	Congenital disorder of glycosylation, type IIp, 616829
TMEM70	98	93,9	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	99,5	97,1	100	99,9	{Autism, susceptibility to, X-linked 6}, 300872
TPI1	99,8	97,5	100	100	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	99,8	99	100	100	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPMT	99,1	90,1	100	100	{Thiopurines, poor metabolism of, 1}, 610460
TPP1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TRAK1	93,3	92,9	100	99,9	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356

TRAPPC2L	100	100	100	100	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TREH	96,9	92,1	100	100	Trehalase deficiency, 612119
TUSC3	100	99,5	100	100	Mental retardation, autosomal recessive 7, 611093
TYMP	100	97	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	99,9	99,6	100	100	No OMIM disease ID
TYR	100	100	100	100	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 Albinism, oculocutaneous, type IA, 203100 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800
TYRP1	100	99,8	100	100	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 Albinism, oculocutaneous, type III, 203290
UGT1A1	100	100	100	100	[Gilbert syndrome], 143500 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 [Bilirubin, serum level of, QTL1], 601816
UMPS	100	99,4	97	97	Orotic aciduria, 258900
UPB1	100	100	100	100	Beta-ureidopropionase deficiency, 613161
UROC1	100	100	100	100	?Urocanase deficiency, 276880
UROD	98,9	96,1	100	100	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100	99,9	100	100	Porphyria, congenital erythropoietic, 263700
VMA21	99	94,6	100	98,6	Myopathy, X-linked, with excessive autophagy, 310440
VPS13B	99,5	98,2	99,5	99,4	Cohen syndrome, 216550
VPS33A	91,3	89,8	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
XDH	100	99,9	100	100	Xanthinuria, type I, 278300
XYLT1	97,4	89,6	98,1	94,8	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777
XYLT2	100	98,3	96,7	96,7	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822
ZBTB11	99,9	99,6	100	100	Intellectual developmental disorder, autosomal recessive 69, 618383

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors