METABOLIC DISORDERS GENE PANEL DG 3.2.0 (703 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
AASS	99,9	99,8	100	99,9	Hyperlysinemia, 238700
ABAT	99,9	97,8	100	100	GABA-transaminase deficiency, 613163
ABCC8	100	99,5	100	100	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCD1	76	72,6	100	100	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	100	99,8	100	100	No OMIM disease ID
ABCD3	99,4	98	100	100	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	99,8	97,7	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	99,9	99,9	100	100	Sitosterolemia 2, 618666
ABCG8	99	97,1	100	100	Sitosterolemia 1, 210250
ABHD12	91,7	86	100	99,8	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100	100	100	100	Chanarin-Dorfman syndrome, 275630
ACACA	98,4	97,9	100	100	No OMIM disease ID
ACAD8	100	100	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	99,8	97,9	100	100	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100	99,4	100	100	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	99,8	97,9	100	100	2-methylbutyrylglycinuria, 610006
ACADVL	99,7	96,6	100	100	VLCAD deficiency, 201475
ACAT1	99,6	97,9	100	99,7	Alpha-methylacetoacetic aciduria, 203750

ACAT2	100	99,9	100	100	No OMIM disease ID
ACBD5	100	98,4	100	99,9	Retinal dystrophy with leukodystrophy, 618863
ACO2	94,1	86,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100	99,3	100	100	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100	99	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACSF3	100	99,5	100	100	Combined malonic and methylmalonic aciduria, 614265
ACSL4	98,3	94,2	100	99,6	Intellectual developmental disorder, X-linked 63, 300387
ACY1	100	99,7	100	100	Aminoacylase 1 deficiency, 609924
ADA	99,7	96,1	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	100	100	100	100	No OMIM disease ID
ADCY5	95,9	92,5	99,2	97,9	Dyskinesia, familial, with facial myokymia, 606703
ADK	83,3	79,7	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	99,2	98,6	100	100	Adenylosuccinase deficiency, 103050
AGA	100	99,9	100	100	Aspartylglucosaminuria, 208400
AGK	90,4	87,9	91,2	91,1	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	99,8	99,5	100	100	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	99	94	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	98,8	95,2	100	99,4	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHCY	99,9	98,8	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	100	100	100	100	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	98,7	95,2	100	99,7	Reticular dysgenesis, 267500
AKR1C1	94,5	87,4	100	100	No OMIM disease ID
AKR1D1	99,8	98,6	100	100	Bile acid synthesis defect, congenital, 2, 235555
ALAD	97,8	92,5	100	100	Porphyria, acute hepatic, 612740
ALAS2	98,7	93,2	100	100	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752

ALDH18A1	100	99,9	100	100	Spastic paraplegia 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603
ALDH1A3	97,6	94,6	100	100	Microphthalmia, isolated 8, 615113
ALDH2	100	100	100	100	Alcohol sensitivity, acute, 610251
ALDH3A2	88,8	88,4	93,2	93,2	Sjogren-Larsson syndrome, 270200
ALDH4A1	100	99,7	100	100	Hyperprolinemia, type II, 239510
ALDH5A1	92,4	83,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100	99,6	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	91,1	84,5	100	100	Epilepsy, pyridoxine-dependent, 266100
ALDOA	98,9	96,3	100	100	Glycogen storage disease XII, 611881
ALDOB	98,8	95,7	100	100	Fructose intolerance, hereditary, 229600
ALG1	53,6	46,9	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100	99,7	100	100	No OMIM disease ID
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100	99,9	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	97,4	90	99,9	99,4	?Congenital disorder of glycosylation, type Is, 300884 Developmental and epileptic encephalopathy 36, 300884
ALG14	100	99,9	100	100	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	100	100	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100	99,5	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,2	93,7	100	99,9	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96,6	95,9	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	99,9	99,3	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALOX12B	100	99,7	100	100	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	100	99,4	100	100	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500

					Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
AMACR	100	100	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	92,5	82,9	100	100	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	100	99,5	100	100	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	100	99,2	100	100	No OMIM disease ID
AMT	100	100	100	100	Glycine encephalopathy, 605899
AP1S1	99,9	99,4	100	100	MEDNIK syndrome, 609313
AP3B2	93,3	89,8	99,9	99	Developmental and epileptic encephalopathy 48, 617276
APOA5	100	99,9	100	99,8	Hyperchylomicronemia, late-onset, 144650
APOC2	100	100	100	100	Hyperlipoproteinemia, type Ib, 207750
APRT	100	100	100	100	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	92,9	92,9	92,9	92,7	Argininemia, 207800
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ARSB	98,8	91	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	99,1	97,3	100	100	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASL	100	99,7	100	100	Argininosuccinic aciduria, 207900
ASNS	98,1	91,2	100	100	Asparagine synthetase deficiency, 615574
ASPA	99,9	99,1	100	100	Canavan disease, 271900
ASS1	93,2	83,2	100	100	Citrullinemia, 215700
ATIC	99,8	99,1	100	100	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A1	100	99,8	100	100	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP6AP1	98,2	93	100	100	Immunodeficiency 47, 300972
ATP6AP2	89,9	69,7	100	99,8	Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A2	99,9	98,7	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200

ATP6V1A	99,8	98,4	100	100	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1E1	92,5	86,1	100	100	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	98,7	96	100	99,9	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8B1	96,6	93,5	100	100	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300
AUH	99,7	99,4	100	99,9	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	100	100	100	100	No OMIM disease ID
B3GALNT2	94,3	89,8	92,5	92,5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	77	73	91,7	81	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GAT3	99,4	96,6	95,4	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	99,7	98,2	100	99,4	Peters-plus syndrome, 261540
B4GALT1	100	99,3	100	100	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	99,7	96,8	100	99,4	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	99,5	97,5	100	100	Hypercholanemia, familial, 607748 Bile acid conjugation defect 1, 619232
BCAT1	100	100	100	100	No OMIM disease ID
BCAT2	100	100	100	100	?Hypervalinemia or hyperleucine-isoleucinemia, 618850
BCKDHA	99,8	97,9	100	100	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,8	95,4	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	99,8	97,8	100	100	Hyperbiliverdinemia, 614156
BMP2	100	100	100	100	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600

BPGM	100	100	100	100	Erythrocytosis, familial, 8, 222800
IMPAD1	100	99,9	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BTD	83	82,9	83,1	83,1	Biotinidase deficiency, 253260
C1GALT1C1	100	99,1	100	100	Tn polyagglutination syndrome, somatic, 300622
CA5A	87,6	85,6	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	99,8	98,5	100	100	Developmental and epileptic encephalopathy 50, 616457
CANT1	100	100	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAT	100	100	100	100	Acatalasemia, 614097
CBLIF	100	99,6	100	100	Intrinsic factor deficiency, 261000
CBS	99,9	98,5	100	100	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CCDC115	95,8	90,2	100	100	Congenital disorder of glycosylation, type IIo, 616828
CD320	100	100	100	99,9	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CEL	90,8	89,2	100	99,9	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	99,3	96,5	100	100	Retinitis pigmentosa 26, 608380
CERS3	99,7	98	100	100	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	99,5	97,9	100	100	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHIT1	99,9	98,2	100	100	No OMIM disease ID
CHKB	100	99,6	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	99,9	98,8	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,9	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100	100	100	100	Macular corneal dystrophy, 217800
CHSY1	97,8	96,3	99,3	96,9	Temtamy preaxial brachydactyly syndrome, 605282
CLCN7	99,4	97,8	100	100	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLN3	92,5	92,4	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69	66,3	71,8	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731

CLN6	99,9	98,9	100	100	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, 4A (Kufs type), autosomal recessive, 204300
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLPB	94,9	94	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CMAS	99,3	96,7	100	100	No OMIM disease ID
COG1	100	99,9	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG2	99,8	98,9	100	100	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100	100	100	100	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	99,1	96,8	100	100	Congenital disorder of glycosylation, type IIi, 613612
COG6	98,5	93,1	100	100	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	100	99,4	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	98,6	95,3	100	100	Congenital disorder of glycosylation, type IIh, 611182
COMT	100	100	100	100	No OMIM disease ID
COQ2	97,6	96,7	97,2	97,2	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	91	89,7	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100	100	100	100	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	99,9	98,5	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,6	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100	99,6	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100	99,2	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	98,7	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
CP	92,6	85,2	100	99,9	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPOX	99,8	97,2	100	100	Coproporphyria, 121300 Harderoporphyria, 618892
CPS1	100	100	100	100	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	99,8	97,6	100	100	CPT deficiency, hepatic, type IA, 255120

CPT2	98,2	97,4	100	100	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CRAT	100	99,9	100	100	?Neurodegeneration with brain iron accumulation 8, 617917
CRPPA	98,4	94,7	100	99,8	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CTH	100	99,9	100	100	Cystathioninuria, 219500
CTNS	100	99,3	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTSA	100	99,6	100	100	Galactosialidosis, 256540
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	100	99,2	100	100	Pycnodysostosis, 265800
CUBN	99,2	97,1	100	100	Imerslund-Grasbeck syndrome 1, 261100
CYB561	92,8	92,7	100	100	Orthostatic hypotension 2, 618182
CYB5R3	99,1	98,1	99,6	98,5	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	99,2	94,5	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	99,9	100	100	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100	99,9	100	100	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	99,9	98,5	100	100	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	98,3	95,7	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	97,4	91,1	100	100	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910

CYP27A1	99,7	98,1	100	100	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100	99,8	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2R1	99,5	96	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	95,3	92	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	98,1	92,7	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99,7	98,2	100	100	D-2-hydroxyglutaric aciduria, 600721
DAO	100	99,9	100	100	No OMIM disease ID
DBH	100	100	100	100	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	99,1	96,1	100	100	Maple syrup urine disease, type II, 248600
DCXR	99,1	94,4	100	100	No OMIM disease ID
DDC	99,2	95	100	100	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	98,5	96,5	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	100	99,8	100	100	?Congenital disorder of glycosylation, type Ir, 614507
DEGS1	100	100	100	100	Leukodystrophy, hypomyelinating, 18, 618404
DGAT1	91,8	87,6	99,9	98,9	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	99,7	98,5	100	100	Nephrotic syndrome, type 7, 615008
DGUOK	99,9	98,8	100	100	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 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	99,4	95,6	95,2	95,2	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DHFR	88,9	76,3	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	100	99,9	100	100	Miller syndrome, 263750
DLD	99,9	99,7	100	99,9	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	99,9	99,7	100	100	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	87,4	87,3	100	100	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	99,3	92,2	100	100	3-methylglutaconic aciduria, type V, 610198

DNM1L	99,6	98,3	100	100	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	98,6	93,9	100	100	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	99,2	98,8	99,9	99,4	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	100	99,9	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	100	99,8	100	100	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPM1	97,4	90,9	98,6	94,6	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	97,7	100	100	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100	100	100	100	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPYD	99,5	96,5	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100	100	100	100	Dihydropyrimidinuria, 222748
DTYMK	100	100	100	100	No OMIM disease ID
EBP	99,5	94,3	100	100	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	100	99,4	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ELOVL1	99,6	96,5	100	100	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	99,7	98,9	100	99,9	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	100	100	100	100	?Glycogen storage disease XIII, 612932
EOGT	79,3	77,8	91,8	88,3	Adams-Oliver syndrome 4, 615297
EPHX1	99,8	97,8	100	100	No OMIM disease ID
EPHX2	99,5	96,5	100	99,9	No OMIM disease ID
ETFA	99,8	99,6	100	99,9	Glutaric acidemia IIA, 231680

ETFB	100	99,9	100	100	Glutaric acidemia IIB, 231680
ETFDH	99,8	99,4	100	100	Glutaric acidemia IIC, 231680
ETHE1	99,3	93,3	100	100	Ethylmalonic encephalopathy, 602473
EXT1	99,6	97,1	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	99,9	99	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EYA1	99,9	99,5	100	100	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
FA2H	92,4	82,6	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FAH	100	99,5	100	99,9	Tyrosinemia, type I, 276700
FAR1	97,4	94	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FBP1	93,6	91,3	93,7	93,7	Fructose-1,6-bisphosphatase deficiency, 229700
FCSK	98	96,1	100	100	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	98,5	96,7	100	100	Squalene synthase deficiency, 618156
FECH	99,9	99,8	100	100	Protoporphyria, erythropoietic, 1, 177000
FH	93,2	87,2	100	100	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FKRP	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	99,8	95,2	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615
FLAD1	100	99,7	100	100	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	99,9	99,7	100	100	Trimethylaminuria, 602079
FOLR1	100	99,9	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	97,7	93,2	100	100	Glutamate formiminotransferase deficiency, 229100
FUCA1	100	100	100	100	Fucosidosis, 230000

FUT2	100	100	100	100	No OMIM disease ID
FUT6	100	100	100	100	No OMIM disease ID
FUT8	99,8	98,9	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	99,1	97,4	100	100	Hemolytic anemia, G6PD deficient (favism), 300908
GAA	100	99,9	100	100	Glycogen storage disease II, 232300
GAD1	100	99,3	100	100	Developmental and epileptic encephalopathy 89, 619124
GALC	99,7	97,6	100	100	Krabbe disease, 245200
GALE	100	100	100	100	Galactose epimerase deficiency, 230350
GALK1	100	99,2	100	100	Galactokinase deficiency with cataracts, 230200
GALM	100	99,5	100	100	Galactosemia IV, 618881
GALNS	100	99,3	100	100	Mucopolysaccharidosis IVA, 253000
GALNT2	99,8	97,1	100	100	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	99,8	98,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100	99,6	100	100	Galactosemia, 230400
GAMT	95	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736
GANAB	99,8	97,8	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 II, 230900 Gaucher disease, type IIIC, 231005 Gaucher disease, type III, 231000 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013
GBA2	100	99,5	100	100	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	100	99,2	100	100	Glutaricaciduria, type I, 231670
GCH1	99,9	97,3	100	100	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910

GCK	95,4	95,4	95,2	92,6	MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	99,4	97,1	100	99,9	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCLM	99,5	95,4	100	100	No OMIM disease ID
GCSH	75,7	64,4	100	100	?Glycine encephalopathy, 605899
GFPT1	99,9	99,4	100	100	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGPS1	99,8	99,8	100	100	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GK	84,2	61,8	100	99,6	Glycerol kinase deficiency, 307030
GLA	91	85,9	91,3	91,3	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	99,2	92,8	100	100	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	88,9	77,8	100	99,9	Glycine encephalopathy, 605899
GLRA1	100	99,8	100	100	Hyperekplexia 1, 149400
GLRX5	97,2	89,6	99,3	95,2	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	96,9	88,5	100	99,9	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Developmental and epileptic encephalopathy 71, 618328
GLUD1	96,4	84,4	100	100	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	73	69	100	100	Glutamine deficiency, congenital, 610015
GLYCTK	98,7	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 (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GMPS	98,2	94,5	100	99,9	No OMIM disease ID
GNE	100	99,5	100	100	Sialuria, 269921 Nonaka myopathy, 605820

GNMT	100	100	100	100	Glycine N-methyltransferase deficiency, 606664
GNPAT	99,5	95,6	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	99,9	99,7	100	100	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	99,8	96,6	100	100	Mucolipidosis III gamma, 252605
GNS	99,2	94,6	100	100	Mucopolysaccharidosis type IIID, 252940
GOT1	100	98,6	100	100	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	94,6	87	100	100	Developmental and epileptic encephalopathy 82, 618721
GPD1	100	99,9	100	100	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100	98,8	100	100	Brugada syndrome 2, 611777
GPHN	99,9	99,1	100	100	Molybdenum cofactor deficiency C, 615501
GPI	100	99,3	100	100	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100	99,9	100	100	Hyperlipoproteinemia, type 1D, 615947
GPT2	99,4	95,3	100	100	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	97,4	88,7	100	100	No OMIM disease ID
GRHPR	83,3	79,2	100	99,3	Hyperoxaluria, primary, type II, 260000
GSS	96,5	96,3	100	100	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GUSB	92,5	90,1	100	100	Mucopolysaccharidosis VII, 253220
GYG1	99,6	97,4	100	100	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100	98	100	100	Glycogen storage disease 0, muscle, 611556
GYS2	99,9	99,4	100	100	Glycogen storage disease 0, liver, 240600
H6PD	99	99	100	100	Cortisone reductase deficiency 1, 604931
HADH	99,2	97,7	100	100	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	95,5	88,3	100	100	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	97,7	87	100	99,9	Trifunctional protein deficiency, 609015
HAGH	100	100	99,1	96,7	No OMIM disease ID

HEXA	93,8	93,1	100	100	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	99,4	96,6	100	100	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	99,9	97,8	100	100	Hemochromatosis, 235200
HGD	100	99,7	100	100	Alkaptonuria, 203500
HGSNAT	86,4	86,2	91,3	89,1	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	93,8	91,3	100	100	No OMIM disease ID
HIBCH	98,2	84,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	100	99,9	100	100	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HMBS	100	98,4	100	100	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HMGCL	100	99,4	100	100	HMG-CoA lyase deficiency, 246450
HMGCS2	100	99,7	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	97,7	90,1	100	100	Heme oxygenase-1 deficiency, 614034
HNF1A	100	99,8	100	100	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700
HNF4A	99,9	98,6	100	100	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850
HOGA1	99,5	95,5	100	100	Hyperoxaluria, primary, type III, 613616
HPD	100	99,8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPDL	100	100	100	100	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	98,6	90,6	99,5	98,4	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	93,6	86,7	100	100	No OMIM disease ID
HSD11B1	100	99,7	100	100	Cortisone reductase deficiency 2, 614662

HSD11B2	87,6	83,8	99,9	97,6	Apparent mineralocorticoid excess, 218030
HSD17B10	99,9	98,3	100	100	HSD10 mitochondrial disease, 300438
HSD17B3	97,8	97,8	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,3	92,8	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	98,9	95	100	100	Bile acid synthesis defect, congenital, 1, 607765
HTRA2	100	99,6	100	100	3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100	100	100	100	?Mucopolysaccharidosis type IX, 601492
IDH2	99,8	97,4	100	100	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	95,4	95,4	100	100	Retinitis pigmentosa 46, 612572
IDI1	99	96,4	100	100	No OMIM disease ID
IDS	99,6	95,3	100	100	Mucopolysaccharidosis II, 309900
IDUA	94,6	87,4	100	100	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IMPDH1	89	81,7	100	99,9	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
INPP5E	96,9	93,2	100	100	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	98,6	94,4	100	99,9	Opsismodysplasia, 258480
INSR	97,3	93	100	99,6	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IREB2	99,9	99,8	100	100	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
ITCH	91,5	90,8	95,3	93,1	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647
IVD	100	99,9	100	100	Isovaleric acidemia, 243500
KCNA2	100	99,6	100	100	Developmental and epileptic encephalopathy 32, 616366
KCNJ11	100	100	100	100	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329

					Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KMT2A	100	99,7	100	99,8	Wiedemann-Steiner syndrome, 605130
KMT2D	99,9	99	100	100	Kabuki syndrome 1, 147920
L2HGDH	98,9	96,4	100	100	L-2-hydroxyglutaric aciduria, 236792
LAMP2	99,3	96	100	99,7	Danon disease, 300257
LARGE1	100	99,7	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	98,8	93,3	100	100	Fish-eye disease, 136120 Norum disease, 245900
LCT	99,6	97,4	100	100	Lactase deficiency, congenital, 223000
LDHA	94,4	89,3	100	100	Glycogen storage disease XI, 612933
LDHB	90,5	77,9	100	100	No OMIM disease ID
LFNG	88,6	86,5	92	87,3	Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	99,8	98,9	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	96,9	94,6	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100	99,4	100	100	Hepatic lipase deficiency, 614025
LIPT1	99,7	99,5	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	98,4	82,4	100	100	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	94,1	89,1	96,1	95,7	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100	99,7	100	100	Lipase deficiency, combined, 246650
LPIN1	99,4	97,2	100	100	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	99,9	99,7	100	100	Majeed syndrome, 609628
LPL	100	100	100	100	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LRAT	100	100	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	76,4	69	100	100	No OMIM disease ID
LYST	99,4	97,8	100	100	Chediak-Higashi syndrome, 214500
MAN1B1	100	99,7	100	100	Rafiq syndrome, 614202

MAN2B1	99,6	97,4	100	100	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	99,9	99,2	100	100	No OMIM disease ID
MANBA	87,1	84,9	100	99,9	Mannosidosis, beta, 248510
MAOA	100	99,8	99,4	97,9	Brunner syndrome, 300615
MAT1A	99,9	98,5	100	100	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBOAT7	100	99,3	100	100	Mental retardation, autosomal recessive 57, 617188
MCCC1	99,9	98,7	100	100	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,9	99,1	100	100	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100	100	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	99,8	98,8	100	100	Mucolipidosis IV, 252650
MDH1	99,7	99,1	100	100	?Developmental and epileptic encephalopathy 88, 618959
MFSD2A	99,5	97,3	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	99,6	99,4	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100	99,9	100	100	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	99,7	99,3	100	99,9	No OMIM disease ID
MLYCD	96,8	92,5	100	99,4	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100	100	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100	99,9	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	91,6	81,3	89,7	89,7	Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Homocystinuria, cblD type, variant 1, 277410
MMUT	99,7	98,2	100	100	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	99,9	97,8	100	100	Xanthinuria, type II, 603592
MOCS1	98,9	95,5	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,4	99,4	100	100	Molybdenum cofactor deficiency B, 252160
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056
MPDU1	100	99,2	100	100	Congenital disorder of glycosylation, type If, 609180

MPI	100	99,5	100	100	Congenital disorder of glycosylation, type Ib, 602579
MRPL44	99,5	97,4	100	100	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS36	94	75,2	100	100	No OMIM disease ID
MSMO1	93,1	86,8	100	100	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	99,9	98,4	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	97,3	95,9	100	100	Homocystinuria due to MTHFR deficiency, 236250
MTM1	98,7	92	100	99,7	Myotubular myopathy, X-linked, 310400
MTMR2	99,5	98,4	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	100	99,9	100	100	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	99,8	98,4	100	100	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MVK	91,4	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
NADK2	99,7	99,3	99,5	96,8	2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	100	100	100	100	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	93,8	91,7	99,9	98,7	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	99,9	97,9	100	100	N-acetylglutamate synthase deficiency, 237310
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NAXD	100	99,9	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100	98,6	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	99,9	99,3	100	100	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NEU1	99,3	96,1	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NGLY1	99,8	99,7	100	100	Congenital disorder of deglycosylation, 615273
NMNAT1	100	99,2	99,4	96,7	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NNT	96,4	96	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736

NPC1	99,9	99	100	100	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100	99,2	100	100	Niemann-pick disease, type C2, 607625
NPL	100	99,3	100	100	No OMIM disease ID
NSD1	100	99,8	100	100	Sotos syndrome 1, 117550
NSDHL	99,8	96,3	100	100	CK syndrome, 300831 CHILD syndrome, 308050
NT5C3A	94,6	82,2	100	100	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100	100	100	100	Calcification of joints and arteries, 211800
NUS1	56,5	42	100	99,9	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	82	73	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	99,4	97,6	100	99,9	Dent disease 2, 300555 Lowe syndrome, 309000
OGDH	100	99,8	100	100	No OMIM disease ID
OPA3	100	99,5	100	100	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	100	99,8	100	100	5-oxoprolinase deficiency, 260005
OTC	100	99,9	100	99,7	Ornithine transcarbamylase deficiency, 311250
OXCT1	99,4	97,6	100	100	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	100	100	100	100	Phenylketonuria, 261600
PANK2	100	99,7	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	99,7	98	100	100	Pyruvate carboxylase deficiency, 266150
PCBD1	100	99,8	100	100	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	98,9	93,4	100	100	Propionicacidemia, 606054
PCCB	96,7	95,4	99	96,2	Propionicacidemia, 606054
PCK1	100	100	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100	100	100	100	No OMIM disease ID
PCYT1A	99,2	95,7	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PCYT2	100	98,3	99,6	97,8	Spastic paraplegia 82, autosomal recessive, 618770
PDSS1	95,2	87,8	97,4	97,4	Coenzyme Q10 deficiency, primary, 2, 614651

PDSS2	98,4	94,3	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	100	99,4	100	100	Prolidase deficiency, 170100
PEX1	99,8	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	98,8	90,6	100	100	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	100	98,3	100	100	Peroxisome biogenesis disorder 14B, 614920
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	95,8	89,4	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,1	93,9	100	100	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	99	94,4	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	99,8	100	100	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	99,4	99,2	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	98,8	100	100	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	96,4	88	100	100	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	88	81	91,3	91,2	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	100	99,7	100	100	Glycogen storage disease VII, 232800
PGAM2	100	100	100	100	Glycogen storage disease X, 261670
PGAP1	98,7	94,6	100	99,8	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	62,6	58,1	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716

PGK1	90,3	73,2	100	100	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94,2	94,1	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PGM3	99,9	99,7	91,7	91,7	Immunodeficiency 23, 615816
PHGDH	99,9	98,2	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	97,8	93,4	100	99,6	Muscle glycogenosis, 300559
PHKA2	100	99,1	100	99,4	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	99,7	99,1	100	100	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	99,8	97,5	100	100	No OMIM disease ID
PHKG2	100	99,9	100	100	Glycogen storage disease IXc, 613027
PHYH	100	98,9	100	100	Refsum disease, 266500
PI4K2A	93,4	87,6	100	100	No OMIM disease ID
PIGA	91,6	82,5	100	99,8	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGB	99,5	97,3	100	100	Developmental and epileptic encephalopathy 80, 618580
PIGC	96	86,2	100	100	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGL	100	99,6	100	100	CHIME syndrome, 280000
PIGM	100	100	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	93,1	89,6	98,8	98,6	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,8	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,6	85,5	100	99,9	Developmental and epileptic encephalopathy 55, 617599
PIGQ	93,4	91,6	100	100	Developmental and epileptic encephalopathy 77, 618548
PIGT	98,1	98	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	99,7	100	100	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	97,7	97,3	100	100	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000

					Colorectal cancer, somatic, 114500 Macrodactyly, somatic, 155500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108
PIK3R1	99,7	98,4	100	100	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	90,9	89,1	99,7	98	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100	99,9	100	100	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	99,9	99,3	100	100	Corneal fleck dystrophy, 121850
PIP5K1C	99,2	96,7	99,9	99,2	Lethal congenital contractural syndrome 3, 611369
PKLR	99,9	98	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PLA2G5	100	100	100	100	No OMIM disease ID
PLA2G6	92,1	90,7	92,3	92,3	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	99,8	99,3	100	100	Platelet-activating factor acetylhydrolase deficiency, 614278
PLCB1	99,9	99,4	100	100	Developmental and epileptic encephalopathy 12, 613722
PLCB4	99,8	98,7	100	100	Auriculocondylar syndrome 2, 614669
PLCD1	99,9	97,3	100	100	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	99,8	98,9	100	100	Nephrotic syndrome, type 3, 610725
PLCG2	100	99,3	100	100	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLIN1	99,6	95,2	100	99,8	Lipodystrophy, familial partial, type 4, 613877
PLOD1	100	98,2	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,2	98,1	100	100	Bruck syndrome 2, 609220
PLOD3	100	98,7	100	100	Lysyl hydroxylase 3 deficiency, 612394

PLPBP	95,1	88,9	100	99,9	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	99,8	99,8	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	99,9	99,6	100	100	?Pancreatic lipase deficiency, 614338
PNMT	99,7	96,8	100	100	No OMIM disease ID
PNP	99,8	98,7	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	99,8	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100	99,8	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
PNPO	99,9	97,1	100	100	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	100	98,9	100	100	Dowling-Degos disease 2, 615327
POGLUT1	99,8	95,9	100	100	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLR3A	99,9	99	100	100	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	99,7	97,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMGNT1	100	99,8	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 (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
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	99,5	97,3	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	99,8	97,3	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
PPCS	100	99,1	100	100	Cardiomyopathy, dilated, 2C, 618189
PPM1K	100	100	100	100	?Maple syrup urine disease, mild variant, 615135

PPOX	99,3	95,7	100	100	Porphyria variegata, 176200
PPT1	90,3	89,9	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	99,4	96,1	99,9	99,3	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKCSH	99,5	94,1	100	100	Polycystic liver disease 1, 174050
PRODH	84	80,2	100	100	Hyperprolinemia, type I, 239500
PRPS1	86,4	86,3	100	99,7	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PSAP	100	99,6	100	100	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PSAT1	92	75,1	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSPH	100	100	100	100	Phosphoserine phosphatase deficiency, 614023
PTEN	99,5	97,2	100	100	Lhermitte-Duclos syndrome, 158350 Cowden syndrome 1, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTGIS	99	95,1	100	100	Hypertension, essential, 145500
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTS	99,5	99	100	99,9	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS3	100	100	100	100	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PYCR1	100	98,2	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,3	100	100	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100	100	100	100	Glycogen storage disease VI, 232700

PYGM	100	100	100	100	McArdle disease, 232600
QDPR	100	98,9	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
RBCK1	99,9	98,3	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	99,3	95,4	100	100	Leber congenital amaurosis 13, 612712
RDH5	100	100	100	100	Fundus albipunctatus, 136880
RFT1	99,7	98,4	100	100	Congenital disorder of glycosylation, type In, 612015
RINT1	99,6	97,6	100	99,9	Infantile liver failure syndrome 3, 618641
RPE65	99,9	98,7	100	100	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100
RPIA	99,1	96,1	100	100	Ribose 5-phosphate isomerase deficiency, 608611
RPN2	100	100	100	100	No OMIM disease ID
RXYLT1	99,2	95,9	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SARDH	93,4	91,6	91,4	91,4	No OMIM disease ID
SAT1	99,7	96,5	100	100	No OMIM disease ID
SC5D	99,9	99,1	100	100	Lathosterolosis, 607330
SCARB2	99,9	99,4	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	99,9	97,9	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	99,9	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	99,9	99,1	100	100	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SELENBP1	100	99,7	100	100	Extraoral halitosis due to MTO deficiency, 618148
SEPSECS	99,9	99,6	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,6	99,5	100	99,9	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	94,8	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SI	99	95,9	100	99,9	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	99,5	98,1	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC12A1	96,2	96	96,2	96,2	Bartter syndrome, type 1, 601678
SLC13A3	99,8	97,9	100	100	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384

SLC16A1	100	98,6	100	100	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	99,6	96,2	100	100	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC18A2	99,9	99,6	100	100	?Parkinsonism-dystonia, infantile, 2, 618049
SLC22A12	100	99,8	100	100	Hypouricemia, renal, 220150
SLC22A5	100	99,6	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A1	96,9	89,8	99,7	98,2	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A13	100	99,4	100	100	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC25A15	99,3	96,6	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	99,9	98	100	100	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100	98,9	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100	99,6	100	100	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A32	100	100	100	99,9	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	97,4	93,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	97,1	94,3	100	100	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC28A1	100	98,3	100	100	No OMIM disease ID
SLC2A1	92,8	92,7	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
SLC2A2	100	99,8	100	100	Fanconi-Bickel syndrome, 227810
SLC2A9	99,3	95	100	100	Hypouricemia, renal, 2, 612076
SLC30A10	100	100	100	100	Hypermanganesemia with dystonia 1, 613280
SLC33A1	99,8	98,5	100	99,8	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	99,7	99,3	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	99,6	97,7	100	100	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	80,4	78,8	81	80,9	?Arthrogryposis, mental retardation, and seizures, 615553

SLC35C1	100	99,4	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	99,6	97,6	100	99,2	Schneckenbecken dysplasia, 269250
SLC37A4	99,8	97,6	100	100	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC39A14	100	99	93,5	93,5	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	99,3	96,3	100	100	Acrodermatitis enteropathica, 201100
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100	99,7	96,6	96,6	Cystinuria, 220100
SLC44A1	98,2	98,1	100	99,9	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC46A1	100	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	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100	99,8	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	100	100	100	100	Renal glucosuria, 233100
SLC6A19	100	100	100	100	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A5	100	99,9	100	100	Hyperekplexia 3, 614618
SLC6A8	94,8	83	99,9	99,5	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100	99,6	100	100	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC7A9	100	99,4	100	100	Cystinuria, 220100
SLCO1B1	98,3	92	100	99,5	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	98,1	88,4	100	99,8	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	100	99,9	100	100	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SMS	87,9	72,1	100	99,5	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNX14	98,9	93,6	100	100	Spinocerebellar ataxia, autosomal recessive 20, 616354

SOD1	100	100	100	100	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SOD2	100	100	100	100	No OMIM disease ID
SPR	100	99,4	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	98,7	93,7	100	100	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100	100	100	99,9	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQOR	100	98	100	100	Sulfide:quinone oxidoreductase deficiency, 619221
SRD5A2	100	98,8	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	100	99,1	100	100	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SSR4	100	99,5	100	100	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	68,8	68,2	95,3	95,2	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	85,9	84	98,7	98,6	Salt and pepper developmental regression syndrome, 609056
STAR	100	99,9	100	100	Lipoid adrenal hyperplasia, 201710
STS	96,8	93,8	97,4	97,2	Ichthyosis, X-linked, 308100
STT3A	100	100	100	100	Congenital disorder of glycosylation, type Iw, 615596
STT3B	99,7	99,4	100	100	?Congenital disorder of glycosylation, type Ix, 615597
STX5	95	89,3	100	100	No OMIM disease ID
SUCLA2	88,8	79,4	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100	99,7	100	99,8	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	91,7	79,1	100	100	No OMIM disease ID
SUGCT	99,6	97,6	100	99,9	Glutaric aciduria III, 231690
SUMF1	98,3	92,5	100	100	Multiple sulfatase deficiency, 272200
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
TAZ	99,3	93,7	100	100	Barth syndrome, 302060
TALDO1	100	98	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

TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	98,5	93,4	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TECR	100	98,5	100	100	Mental retardation, autosomal recessive 14, 614020
TH	99,8	98	100	100	Segawa syndrome, recessive, 605407
TIMM50	98,4	95	100	100	3-methylglutaconic aciduria, type IX, 617698
TK2	99	96	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100	99,8	100	100	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98,6	96,8	98,7	98,7	Short stature, developmental delay, and congenital heart defects, 617044
TMEM106B	99,3	98,3	100	99,9	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	100	100	100	100	Congenital disorder of glycosylation, type IIp, 616829
TMEM70	98,4	94,6	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	98,6	94,1	100	99,7	No OMIM disease ID
TPI1	99,8	98	100	100	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	99,5	97,2	100	100	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPMT	98,4	82	100	100	No OMIM disease ID
TPP1	100	100	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAK1	93,3	93,1	100	99,9	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	99,7	98,7	100	99,9	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC2L	100	100	100	100	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TREH	97,7	93	100	100	Trehalase deficiency, 612119
TUSC3	100	99,7	100	100	Mental retardation, autosomal recessive 7, 611093
TYMP	100	99,4	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	100	99,7	100	100	No OMIM disease ID
TYR	100	100	100	100	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100

TYRP1	100	99,9	100	100	Albinism, oculocutaneous, type III, 203290
UGT1A1	100	100	100	100	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
UMPS	99,9	98,7	97	97	Orotic aciduria, 258900
UPB1	100	100	100	100	Beta-ureidopropionase deficiency, 613161
UROC1	100	99,9	100	100	?Urocanase deficiency, 276880
UROD	98,5	95,5	100	100	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100	99,9	100	100	Porphyria, congenital erythropoietic, 263700
VMA21	99	92,6	100	99,8	Myopathy, X-linked, with excessive autophagy, 310440
VPS13B	99,4	97,8	99,4	99,3	Cohen syndrome, 216550
VPS33A	91,9	89,9	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
XDH	100	99,8	100	100	Xanthinuria, type I, 278300
XYLT1	97,8	91,1	97,7	94,1	Desbuquois dysplasia 2, 615777
XYLT2	99,9	97,1	96,7	96,7	Spondyloocular syndrome, 605822
ZBTB11	99,9	99,3	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.
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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors