METABOLIC DISORDERS GENE PANEL DG 3.4.0 (723 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AASS	100,0%	100,0%	Hyperlysinemia, 238700
ABAT	100,0%	100,0%	GABA-transaminase deficiency, 613163
ABCC8	100,0%	100,0%	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	100,0%	100,0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	100,0%	100,0%	No OMIM Disease ID
ABCD3	100,0%	100,0%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100,0%	100,0%	Sitosterolemia 2, 618666
ABCG8	100,0%	100,0%	Sitosterolemia 1, 210250
ABHD12	100,0%	100,0%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100,0%	100,0%	Chanarin-Dorfman syndrome, 275630
ACACA	100,0%	100,0%	No OMIM Disease ID
ACAD8	100,0%	100,0%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	100,0%	100,0%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100,0%	100,0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100,0%	100,0%	2-methylbutyrylglycinuria, 610006
ACADVL	100,0%	100,0%	VLCAD deficiency, 201475
ACAT1	100,0%	100,0%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	100,0%	100,0%	No OMIM Disease ID
ACBD5	100,0%	100,0%	Retinal dystrophy with leukodystrophy, 618863
ACO2	100,0%	100,0%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559

ACOX1	100,0%	100,0%	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100,0%	100,0%	Bile acid synthesis defect, congenital, 6, 617308
ACSF3	100,0%	100,0%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100,0%	100,0%	Intellectual developmental disorder, X-linked 63, 300387
ACY1	100,0%	100,0%	Aminoacylase 1 deficiency, 609924
ADA	100,0%	100,0%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	100,0%	100,0%	No OMIM Disease ID
ADCY5	100,0%	99,9%	Dyskinesia with orofacial involvement, autosomal dominant, 606703 Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADK	84,5%	84,5%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	100,0%	100,0%	Adenylosuccinase deficiency, 103050
AGA	100,0%	100,0%	Aspartylglucosaminuria, 208400
AGK	91,2%	91,2%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	100,0%	100,0%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	100,0%	99,9%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	100,0%	100,0%	Hyperoxaluria, primary, type 1, 259900
AHCY	100,0%	100,0%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AK1	100,0%	100,0%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	100,0%	100,0%	Reticular dysgenesis, 267500
AKR1C1	100,0%	100,0%	No OMIM Disease ID
AKR1D1	100,0%	100,0%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	100,0%	100,0%	Porphyria, acute hepatic, 612740
ALAS2	100,0%	100,0%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	100,0%	100,0%	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	100,0%	100,0%	Microphthalmia, isolated 8, 615113
ALDH2	100,0%	100,0%	Alcohol sensitivity, acute, 610251
ALDH3A2	93,2%	93,2%	Sjogren-Larsson syndrome, 270200

ALDH4A1	100,0%	100,0%	Hyperprolinemia, type II, 239510
ALDH5A1	100,0%	100,0%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100,0%	100,0%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	100,0%	100,0%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	100,0%	100,0%	Glycogen storage disease XII, 611881
ALDOB	100,0%	100,0%	Fructose intolerance, hereditary, 229600
ALG1	100,0%	100,0%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100,0%	100,0%	No OMIM Disease ID
ALG11	96,8%	96,8%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100,0%	100,0%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100,0%	99,9%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100,0%	100,0%	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	100,0%	100,0%	Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100,0%	100,0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100,0%	100,0%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96,6%	96,6%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100,0%	100,0%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALOX12B	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	100,0%	100,0%	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
AMACR	100,0%	100,0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMN	100,0%	100,0%	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	100,0%	100,0%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	100,0%	100,0%	No OMIM Disease ID
AMT	100,0%	100,0%	Glycine encephalopathy, 605899
AP1S1	100,0%	100,0%	MEDNIK syndrome, 609313
AP3B2	100,0%	99,7%	Developmental and epileptic encephalopathy 48, 617276
APOA5	100,0%	100,0%	Hyperchylomicronemia, late-onset, 144650
APOC2	100,0%	100,0%	Hyperlipoproteinemia, type Ib, 207750

APRT	100,0%	100,0%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	92,9%	92,9%	Argininemia, 207800
ARSA	100,0%	100,0%	Metachromatic leukodystrophy, 250100
ARSB	100,0%	100,0%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ASAH1	100,0%	100,0%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASL	100,0%	100,0%	Argininosuccinic aciduria, 207900
ASNS	100,0%	100,0%	Asparagine synthetase deficiency, 615574
ASPA	100,0%	100,0%	Canavan disease, 271900
ASS1	100,0%	100,0%	Citrullinemia, 215700
ATIC	100,0%	100,0%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A1	100,0%	100,0%	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP6AP1	100,0%	100,0%	Immunodeficiency 47, 300972
ATP6AP2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A2	100,0%	100,0%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	100,0%	100,0%	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1E1	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
ATP7B	100,0%	100,0%	Wilson disease, 277900
ATP8B1	100,0%	100,0%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300
AUH	100,0%	100,0%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	100,0%	100,0%	No OMIM Disease ID
B3GALNT2	92,5%	92,5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	99,8%	98,8%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GAT3	96,2%	94,9%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	100,0%	100,0%	Peters-plus syndrome, 261540

B4GALT1	100,0%	100,0%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	100,0%	100,0%	Bile acid conjugation defect 1, 619232
BCAT1	100,0%	100,0%	No OMIM Disease ID
BCAT2	100,0%	100,0%	?Hypervalinemia or hyperleucine-isoleucinemia, 618850
BCKDHA	100,0%	100,0%	Maple syrup urine disease, type Ia, 248600
BCKDHB	100,0%	100,0%	Maple syrup urine disease, type Ib, 248600
BCKDK	100,0%	100,0%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
BCO1	100,0%	100,0%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BLVRA	100,0%	100,0%	Hyperbiliverdinemia, 614156
BMP2	100,0%	100,0%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BPGM	100,0%	100,0%	Erythrocytosis, familial, 8, 222800
IMPAD1	100,0%	100,0%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BSCL2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	83,1%	83,1%	Biotinidase deficiency, 253260
C1GALT1C1	100,0%	100,0%	Tn polyagglutination syndrome, somatic, 300622
C2orf69	100,0%	100,0%	Combined oxidative phosphorylation deficiency 53, 619423
CA5A	87,7%	87,7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	100,0%	100,0%	Developmental and epileptic encephalopathy 50, 616457
CANT1	100,0%	100,0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAT	100,0%	100,0%	Acatalasemia, 614097
CAV1	100,0%	100,0%	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAVIN1	100,0%	100,0%	Lipodystrophy, congenital generalized, type 4, 613327
CBLIF	100,0%	100,0%	Intrinsic factor deficiency, 261000
CBS	100,0%	100,0%	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CCDC115	100,0%	100,0%	Congenital disorder of glycosylation, type IIo, 616828
CD320	100,0%	100,0%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CEL	100,0%	99,9%	Maturity-onset diabetes of the young, type VIII, 609812

CERKL	100,0%	100,0%	Retinitis pigmentosa 26, 608380
CERS3	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	100,0%	100,0%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHIT1	100,0%	100,0%	No OMIM Disease ID
CHKB	100,0%	100,0%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100,0%	100,0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100,0%	100,0%	Macular corneal dystrophy, 217800
CHSY1	100,0%	99,9%	Temtamy preaxial brachydactyly syndrome, 605282
CIDEC	100,0%	100,0%	?Lipodystrophy, familial partial, type 5, 615238
CLCN7	100,0%	100,0%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLN3	92,7%	92,5%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	71,7%	71,6%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLPB	100,0%	100,0%	Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CMAS	100,0%	100,0%	No OMIM Disease ID
COG1	100,0%	100,0%	Congenital disorder of glycosylation, type IIg, 611209
COG2	100,0%	100,0%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100,0%	100,0%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	100,0%	100,0%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100,0%	100,0%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	100,0%	100,0%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100,0%	100,0%	Congenital disorder of glycosylation, type IIh, 611182
COMT	100,0%	100,0%	No OMIM Disease ID
COQ2	97,2%	97,2%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 7, 616276

COQ5	100,0%	100,0%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100,0%	100,0%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100,0%	100,0%	Nephrotic syndrome, type 9, 615573
COQ9	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	100,0%	100,0%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPOX	100,0%	100,0%	Coproporphyria, 121300 Harderoporphyria, 618892
CPS1	100,0%	100,0%	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	100,0%	100,0%	CPT deficiency, hepatic, type IA, 255120
CPT2	100,0%	100,0%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CRAT	100,0%	100,0%	?Neurodegeneration with brain iron accumulation 8, 617917
CRPPA	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CTH	100,0%	100,0%	Cystathioninuria, 219500
CTNS	100,0%	100,0%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTSA	100,0%	100,0%	Galactosialidosis, 256540
CTSC	100,0%	100,0%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CTSD	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSK	100,0%	100,0%	Pycnodysostosis, 265800
CUBN	100,0%	100,0%	Imerslund-Grasbeck syndrome 1, 261100
CYB561	100,0%	100,0%	Orthostatic hypotension 2, 618182
CYB5R3	100,0%	100,0%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYP11A1	100,0%	100,0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100,0%	100,0%	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010

CYP11B2	100,0%	100,0%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	100,0%	100,0%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100,0%	100,0%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	100,0%	100,0%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	100,0%	100,0%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP27A1	100,0%	100,0%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100,0%	100,0%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	100,0%	100,0%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	100,0%	100,0%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	100,0%	100,0%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	100,0%	100,0%	D-2-hydroxyglutaric aciduria, 600721
DAO	100,0%	100,0%	No OMIM Disease ID
DBH	100,0%	100,0%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	100,0%	100,0%	Maple syrup urine disease, type II, 248600
DCXR	100,0%	100,0%	No OMIM Disease ID
DDC	100,0%	100,0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100,0%	100,0%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	100,0%	100,0%	?Congenital disorder of glycosylation, type Ir, 614507
DEGS1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 18, 618404
DGAT1	100,0%	100,0%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	100,0%	100,0%	Nephrotic syndrome, type 7, 615008
DGUOK	100,0%	100,0%	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%	Desmosterolosis, 602398
DHCR7	100,0%	100,0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	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	100,0%	100,0%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839

DHODH	100,0%	100,0%	Miller syndrome, 263750
DLD	100,0%	100,0%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	100,0%	100,0%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	100,0%	100,0%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100,0%	100,0%	3-methylglutaconic aciduria, type V, 610198
DNM1L	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	100,0%	100,0%	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	100,0%	99,7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOLK	100,0%	100,0%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	100,0%	100,0%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPM1	99,8%	97,8%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100,0%	100,0%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100,0%	100,0%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPYD	100,0%	100,0%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100,0%	100,0%	Dihydropyrimidinuria, 222748
DTYMK	100,0%	100,0%	No OMIM Disease ID
EBP	100,0%	100,0%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	100,0%	100,0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDEM3	100,0%	100,0%	Congenital disorder of glycosylation, type 2V, 619493
ELOVL1	100,0%	100,0%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100,0%	100,0%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENO3	100,0%	100,0%	Glycogen storage disease XIII, 612932
EOGT	94,3%	90,6%	Adams-Oliver syndrome 4, 615297
EPG5	100,0%	100,0%	Vici syndrome, 242840

EPHX1	100,0%	100,0%	No OMIM Disease ID
EPHX2	100,0%	100,0%	No OMIM Disease ID
ETFA	100,0%	100,0%	Glutaric acidemia IIA, 231680
ETFB	100,0%	100,0%	Glutaric acidemia IIB, 231680
ETFDH	100,0%	100,0%	Glutaric acidemia IIC, 231680
ETHE1	100,0%	100,0%	Ethylmalonic encephalopathy, 602473
EXT1	100,0%	100,0%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100,0%	100,0%	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EYA1	100,0%	100,0%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
FA2H	100,0%	100,0%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	100,0%	100,0%	Tyrosinemia, type I, 276700
FAR1	100,0%	100,0%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FBN1	100,0%	100,0%	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900
FBP1	93,7%	93,7%	Fructose-1,6-bisphosphatase deficiency, 229700
FBP2	100,0%	100,0%	No OMIM Disease ID
FCSK	100,0%	100,0%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	100,0%	100,0%	Squalene synthase deficiency, 618156
FECH	100,0%	100,0%	Protoporphyria, erythropoietic, 1, 177000
FH	100,0%	100,0%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FKRP	100,0%	100,0%	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	100,0%	100,0%	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	100,0%	100,0%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FMO3	100,0%	100,0%	Trimethylaminuria, 602079
FOLR1	100,0%	100,0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	100,0%	100,0%	Glutamate formiminotransferase deficiency, 229100
FUCA1	100,0%	100,0%	Fucosidosis, 230000
FUT2	100,0%	100,0%	No OMIM Disease ID
FUT6	100,0%	100,0%	No OMIM Disease ID
FUT8	100,0%	100,0%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
G6PC	100,0%	100,0%	Glycogen storage disease Ia, 232200
G6PC3	100,0%	100,0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	100,0%	100,0%	Hemolytic anemia, G6PD deficient (favism), 300908
GAA	100,0%	100,0%	Glycogen storage disease II, 232300
GAD1	100,0%	100,0%	Developmental and epileptic encephalopathy 89, 619124
GALC	100,0%	100,0%	Krabbe disease, 245200
GALE	100,0%	100,0%	Galactose epimerase deficiency, 230350
GALK1	100,0%	100,0%	Galactokinase deficiency with cataracts, 230200
GALM	100,0%	100,0%	Galactosemia IV, 618881
GALNS	100,0%	100,0%	Mucopolysaccharidosis IVA, 253000
GALNT2	100,0%	100,0%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100,0%	100,0%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100,0%	100,0%	Galactosemia, 230400
GAMT	100,0%	100,0%	Cerebral creatine deficiency syndrome 2, 612736
GANAB	100,0%	100,0%	Polycystic kidney disease 3, 600666
GATM	100,0%	100,0%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBA	100,0%	100,0%	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	100,0%	100,0%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100,0%	100,0%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	100,0%	100,0%	Glutaricaciduria, type I, 231670

GCH1	100,0%	100,0%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	97,0%	93,1%	MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	100,0%	100,0%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCLM	100,0%	100,0%	No OMIM Disease ID
GCSH	100,0%	100,0%	?Glycine encephalopathy, 605899
GFPT1	100,0%	100,0%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGPS1	100,0%	100,0%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GK	100,0%	100,0%	Glycerol kinase deficiency, 307030
GLA	91,3%	91,3%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	100,0%	100,0%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	100,0%	100,0%	Glycine encephalopathy, 605899
GLRA1	100,0%	100,0%	Hyperekplexia 1, 149400
GLRX5	100,0%	100,0%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	100,0%	100,0%	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	100,0%	100,0%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100,0%	100,0%	Glutamine deficiency, congenital, 610015
GLYCTK	100,0%	100,0%	D-glyceric aciduria, 220120
GM2A	100,0%	100,0%	GM2-gangliosidosis, AB variant, 272750
GMPPA	100,0%	100,0%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100,0%	100,0%	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	100,0%	100,0%	No OMIM Disease ID
GNE	100,0%	100,0%	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100,0%	100,0%	Glycine N-methyltransferase deficiency, 606664

GNPAT	100,0%	100,0%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100,0%	100,0%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	100,0%	100,0%	Mucolipidosis III gamma, 252605
GNS	100,0%	100,0%	Mucopolysaccharidosis type IIID, 252940
GOT1	100,0%	100,0%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	100,0%	100,0%	Developmental and epileptic encephalopathy 82, 618721
GPD1	100,0%	100,0%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100,0%	100,0%	Brugada syndrome 2, 611777
GPHN	100,0%	100,0%	Molybdenum cofactor deficiency C, 615501
GPI	100,0%	100,0%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100,0%	100,0%	Hyperlipoproteinemia, type 1D, 615947
GPT2	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	100,0%	100,0%	No OMIM Disease ID
GRHPR	100,0%	99,9%	Hyperoxaluria, primary, type II, 260000
GSS	100,0%	100,0%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GUSB	100,0%	100,0%	Mucopolysaccharidosis VII, 253220
GYG1	100,0%	100,0%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100,0%	100,0%	Glycogen storage disease 0, muscle, 611556
GYS2	100,0%	100,0%	Glycogen storage disease 0, liver, 240600
H6PD	100,0%	100,0%	Cortisone reductase deficiency 1, 604931
HADH	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	100,0%	100,0%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	100,0%	100,0%	Trifunctional protein deficiency, 609015
HAGH	100,0%	99,7%	No OMIM Disease ID
HEXA	100,0%	100,0%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	100,0%	100,0%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	100,0%	100,0%	Hemochromatosis, 235200
HGD	100,0%	100,0%	Alkaptonuria, 203500

HGSNAT	92,1%	92,1%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	100,0%	100,0%	No OMIM Disease ID
HIBCH	100,0%	100,0%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	100,0%	100,0%	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	100,0%	100,0%	Holocarboxylase synthetase deficiency, 253270
HMBS	100,0%	100,0%	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HMGCL	100,0%	100,0%	HMG-CoA lyase deficiency, 246450
HMGCS2	100,0%	100,0%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	100,0%	100,0%	Heme oxygenase-1 deficiency, 614034
HNF1A	100,0%	100,0%	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700
HNF4A	100,0%	100,0%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850
HOGA1	100,0%	100,0%	Hyperoxaluria, primary, type III, 613616
HPD	100,0%	100,0%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPDL	100,0%	100,0%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	100,0%	100,0%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HS6ST1	100,0%	100,0%	No OMIM Disease ID
HSD11B1	100,0%	100,0%	Cortisone reductase deficiency 2, 614662
HSD11B2	100,0%	100,0%	Apparent mineralocorticoid excess, 218030
HSD17B10	100,0%	100,0%	HSD10 mitochondrial disease, 300438
HSD17B3	99,0%	99,0%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100,0%	100,0%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	100,0%	100,0%	Bile acid synthesis defect, congenital, 1, 607765
HTRA2	100,0%	100,0%	3-methylglutaconic aciduria, type VIII, 617248

HYAL1	100,0%	100,0%	Mucopolysaccharidosis type IX, 601492
IDH2	100,0%	100,0%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	100,0%	100,0%	Retinitis pigmentosa 46, 612572
IDI1	100,0%	100,0%	No OMIM Disease ID
IDS	100,0%	100,0%	Mucopolysaccharidosis II, 309900
IDUA	100,0%	100,0%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IMPDH1	100,0%	100,0%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
INPP5E	100,0%	100,0%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	100,0%	100,0%	Opsismodysplasia, 258480
INSR	100,0%	100,0%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IREB2	100,0%	100,0%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
ITCH	95,6%	93,9%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITPA	100,0%	100,0%	Developmental and epileptic encephalopathy 35, 616647
IVD	100,0%	100,0%	Isovaleric acidemia, 243500
KCNA2	100,0%	100,0%	Developmental and epileptic encephalopathy 32, 616366
KCNJ11	100,0%	100,0%	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	100,0%	100,0%	Wiedemann-Steiner syndrome, 605130
KMT2D	100,0%	100,0%	Kabuki syndrome 1, 147920
L2HGDH	100,0%	100,0%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	100,0%	100,0%	Danon disease, 300257
LARGE1	100,0%	100,0%	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	100,0%	100,0%	Fish-eye disease, 136120 Norum disease, 245900
LCT	100,0%	100,0%	Lactase deficiency, congenital, 223000
LDHA	100,0%	100,0%	Glycogen storage disease XI, 612933
LDHB	100,0%	100,0%	No OMIM Disease ID

LFNG	99,3%	96,6%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	100,0%	100,0%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	95,2%	95,2%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100,0%	100,0%	Hepatic lipase deficiency, 614025
LIPE	100,0%	100,0%	Lipodystrophy, familial partial, type 6, 615980
LIPT1	100,0%	100,0%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100,0%	100,0%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	96,1%	96,1%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100,0%	100,0%	Lipase deficiency, combined, 246650
LMNA	100,0%	100,0%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMNB2	100,0%	99,8%	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LPIN1	100,0%	100,0%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	100,0%	100,0%	Majeed syndrome, 609628
LPL	100,0%	100,0%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LRAT	100,0%	100,0%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LTC4S	100,0%	100,0%	No OMIM Disease ID
LYST	100,0%	100,0%	Chediak-Higashi syndrome, 214500
MAN1B1	100,0%	100,0%	Rafiq syndrome, 614202
MAN2B1	100,0%	100,0%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100,0%	100,0%	No OMIM Disease ID
MANBA	100,0%	100,0%	Mannosidosis, beta, 248510
MAOA	99,9%	99,4%	Brunner syndrome, 300615

MAT1A	100,0%	100,0%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBOAT7	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 57, 617188
MCCC1	100,0%	100,0%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	100,0%	100,0%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100,0%	100,0%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	100,0%	100,0%	Mucolipidosis IV, 252650
MDH1	100,0%	100,0%	?Developmental and epileptic encephalopathy 88, 618959
MFSD2A	100,0%	100,0%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100,0%	100,0%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100,0%	100,0%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	100,0%	100,0%	Pontocerebellar hypoplasia, type 16, 619527
MLYCD	100,0%	100,0%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100,0%	100,0%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100,0%	100,0%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	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	100,0%	100,0%	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	100,0%	100,0%	Xanthinuria, type II, 603592
MOCS1	100,0%	100,0%	Molybdenum cofactor deficiency A, 252150
MOCS2	100,0%	100,0%	Molybdenum cofactor deficiency B, 252160
MOGS	100,0%	100,0%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPDU1	100,0%	100,0%	Congenital disorder of glycosylation, type If, 609180
MPI	100,0%	100,0%	Congenital disorder of glycosylation, type Ib, 602579
MRPL44	100,0%	100,0%	Combined oxidative phosphorylation deficiency 16, 615395
MRPS36	100,0%	100,0%	No OMIM Disease ID
MSMO1	100,0%	100,0%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	100,0%	100,0%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	100,0%	100,0%	Homocystinuria due to MTHFR deficiency, 236250
MTM1	100,0%	100,0%	Myopathy, centronuclear, X-linked, 310400
MTMR2	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940

MTRR	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MVK	90,5%	90,5%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
NADK2	100,0%	100,0%	2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	100,0%	100,0%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	100,0%	100,0%	N-acetylglutamate synthase deficiency, 237310
NANS	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NAXD	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100,0%	100,0%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NEU1	100,0%	100,0%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NGLY1	100,0%	100,0%	Congenital disorder of deglycosylation 1, 615273
NMNAT1	100,0%	98,5%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NNT	96,4%	96,4%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NPC1	100,0%	100,0%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100,0%	100,0%	Niemann-pick disease, type C2, 607625
NPL	100,0%	100,0%	No OMIM Disease ID
NSD1	100,0%	100,0%	Sotos syndrome, 117550
NSDHL	100,0%	100,0%	CK syndrome, 300831 CHILD syndrome, 308050
NT5C3A	100,0%	100,0%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100,0%	100,0%	Calcification of joints and arteries, 211800
NUS1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OAT	100,0%	100,0%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	100,0%	100,0%	Dent disease 2, 300555 Lowe syndrome, 309000

ODC1	100,0%	100,0%	Bachmann-Bupp syndrome, 619075
OGDH	100,0%	100,0%	?Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100,0%	100,0%	Yoon-Bellen neurodevelopmental syndrome, 619701
OPA3	100,0%	100,0%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	100,0%	100,0%	5-oxoprolinase deficiency, 260005
OTC	100,0%	100,0%	Ornithine transcarbamylase deficiency, 311250
OXCT1	100,0%	100,0%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	100,0%	100,0%	Phenylketonuria, 261600
PANK2	100,0%	100,0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PC	100,0%	100,0%	Pyruvate carboxylase deficiency, 266150
PCBD1	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	100,0%	100,0%	Propionicacidemia, 606054
PCCB	99,9%	98,1%	Propionicacidemia, 606054
PCK1	100,0%	100,0%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100,0%	100,0%	No OMIM Disease ID
PCYT1A	100,0%	100,0%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PCYT2	100,0%	100,0%	Spastic paraplegia 82, autosomal recessive, 618770
PDSS1	97,4%	97,4%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	100,0%	100,0%	Prolidase deficiency, 170100
PEX1	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	100,0%	100,0%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100,0%	100,0%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100,0%	100,0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	100,0%	100,0%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100,0%	100,0%	Peroxisome biogenesis disorder 12A (Zellweger), 614886

PEX2	100,0%	100,0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100,0%	100,0%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100,0%	100,0%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	100,0%	100,0%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100,0%	100,0%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	100,0%	100,0%	Glycogen storage disease VII, 232800
PGAM2	100,0%	100,0%	Glycogen storage disease X, 261670
PGAP1	100,0%	100,0%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	100,0%	100,0%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94,2%	94,2%	Congenital disorder of glycosylation, type It, 614921
PGM2L1	100,0%	100,0%	No OMIM Disease ID
PGM3	91,7%	91,7%	Immunodeficiency 23, 615816
PHGDH	100,0%	100,0%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	100,0%	99,9%	Muscle glycogenosis, 300559
PHKA2	100,0%	100,0%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	100,0%	100,0%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	100,0%	100,0%	No OMIM Disease ID
PHKG2	100,0%	100,0%	Glycogen storage disease IXc, 613027
PHYH	100,0%	100,0%	Refsum disease, 266500
PI4K2A	100,0%	100,0%	No OMIM Disease ID
PIGA	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100,0%	100,0%	Developmental and epileptic encephalopathy 80, 618580

PIGC	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGL	100,0%	100,0%	CHIME syndrome, 280000
PIGM	100,0%	100,0%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	98,8%	98,8%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	100,0%	100,0%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100,0%	100,0%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGT	100,0%	100,0%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100,0%	100,0%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	100,0%	100,0%	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 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 Macrodactyly, somatic,,
PIK3R1	100,0%	100,0%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100,0%	100,0%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	100,0%	100,0%	Corneal fleck dystrophy, 121850
PIP5K1C	100,0%	100,0%	Lethal congenital contractural syndrome 3, 611369
PKLR	100,0%	100,0%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PLA2G5	100,0%	100,0%	No OMIM Disease ID
PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600

PLA2G7	100,0%	100,0%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLCB1	100,0%	100,0%	Developmental and epileptic encephalopathy 12, 613722
PLCB4	100,0%	100,0%	Auriculocondylar syndrome 2, 614669
PLCD1	100,0%	100,0%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	100,0%	100,0%	Nephrotic syndrome, type 3, 610725
PLCG2	100,0%	100,0%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLIN1	100,0%	100,0%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	100,0%	100,0%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100,0%	100,0%	Bruck syndrome 2, 609220
PLOD3	100,0%	100,0%	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	100,0%	100,0%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	100,0%	100,0%	?Pancreatic lipase deficiency, 614338
PNMT	100,0%	100,0%	No OMIM Disease ID
PNP	100,0%	100,0%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	100,0%	100,0%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100,0%	100,0%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
PNPO	100,0%	100,0%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	100,0%	100,0%	Dowling-Degos disease 2, 615327
POGLUT1	100,0%	100,0%	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLD1	100,0%	100,0%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLR3A	100,0%	100,0%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POMGNT1	100,0%	100,0%	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	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830

POMK	100,0%	100,0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	100,0%	100,0%	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	100,0%	100,0%	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
PPARG	98,3%	98,3%	Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 Carotid intimal medial thickness 1, 609338
PPCS	100,0%	100,0%	Cardiomyopathy, dilated, 2C, 618189
PPM1K	100,0%	100,0%	?Maple syrup urine disease, mild variant, 615135
PPOX	100,0%	100,0%	Porphyria variegata, 176200
PPT1	82,5%	82,5%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	100,0%	99,9%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKCSH	100,0%	100,0%	Polycystic liver disease 1, 174050
PRODH	100,0%	100,0%	Hyperprolinemia, type I, 239500
PRPS1	100,0%	100,0%	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	100,0%	100,0%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PSAT1	100,0%	100,0%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSPH	100,0%	100,0%	Phosphoserine phosphatase deficiency, 614023
PTEN	100,0%	100,0%	Cowden syndrome 1, 158350 Lhermitte-Duclos disease, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTGIS	100,0%	100,0%	Hypertension, essential, 145500

PTPN11	100,0%	100,0%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTS	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS3	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PYCR1	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100,0%	100,0%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100,0%	100,0%	Glycogen storage disease VI, 232700
PYGM	100,0%	100,0%	McArdle disease, 232600
QDPR	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RBCK1	100,0%	100,0%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	100,0%	100,0%	Leber congenital amaurosis 13, 612712
RDH5	100,0%	100,0%	Fundus albipunctatus, 136880
RFT1	100,0%	100,0%	Congenital disorder of glycosylation, type In, 612015
RINT1	100,0%	100,0%	Infantile liver failure syndrome 3, 618641
RPE65	100,0%	100,0%	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100
RPIA	100,0%	100,0%	Ribose 5-phosphate isomerase deficiency, 608611
RPN2	100,0%	100,0%	No OMIM Disease ID
RXYLT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SARDH	91,4%	91,4%	No OMIM Disease ID
SAT1	100,0%	100,0%	No OMIM Disease ID
SC5D	100,0%	100,0%	Lathosterolosis, 607330
SCARB2	100,0%	100,0%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	100,0%	100,0%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	100,0%	100,0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SELENBP1	100,0%	100,0%	Extraoral halitosis due to MTO deficiency, 618148
SEPSECS	100,0%	100,0%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGSH	100,0%	100,0%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SI	100,0%	100,0%	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	100,0%	100,0%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363

SLC12A1	96,2%	96,2%	Bartter syndrome, type 1, 601678
SLC13A3	100,0%	100,0%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC16A1	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	100,0%	100,0%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC18A2	100,0%	100,0%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC22A12	100,0%	100,0%	Hypouricemia, renal, 220150
SLC22A5	100,0%	100,0%	Carnitine deficiency, systemic primary, 212140
SLC25A1	100,0%	100,0%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A13	100,0%	100,0%	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC25A15	100,0%	100,0%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100,0%	100,0%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100,0%	100,0%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A32	100,0%	100,0%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	100,0%	100,0%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	100,0%	100,0%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC28A1	100,0%	100,0%	No OMIM Disease ID
SLC2A1	100,0%	100,0%	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	100,0%	100,0%	Fanconi-Bickel syndrome, 227810
SLC2A9	100,0%	100,0%	Hypouricemia, renal, 2, 612076
SLC30A10	100,0%	100,0%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	100,0%	100,0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A1	100,0%	100,0%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100,0%	100,0%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	81,0%	81,0%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35C1	100,0%	100,0%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100,0%	100,0%	Schneckenbecken dysplasia, 269250

SLC37A4	100,0%	100,0%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC39A14	93,6%	93,5%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	100,0%	100,0%	Acrodermatitis enteropathica, 201100
SLC39A8	100,0%	100,0%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	96,6%	96,6%	Cystinuria, 220100
SLC44A1	100,0%	100,0%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC46A1	100,0%	100,0%	Folate malabsorption, hereditary, 229050
SLC52A1	100,0%	100,0%	Riboflavin deficiency, 615026
SLC52A2	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100,0%	100,0%	Glucose/galactose malabsorption, 606824
SLC5A2	100,0%	100,0%	Renal glucosuria, 233100
SLC6A19	100,0%	100,0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A5	100,0%	100,0%	Hyperekplexia 3, 614618
SLC6A8	100,0%	100,0%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100,0%	100,0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100,0%	100,0%	Lysinuric protein intolerance, 222700
SLC7A9	100,0%	100,0%	Cystinuria, 220100
SLCO1B1	99,9%	99,9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	100,0%	100,0%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	100,0%	100,0%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SMS	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNX14	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOD1	100,0%	100,0%	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SOD2	100,0%	100,0%	No OMIM Disease ID
SPR	100,0%	100,0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQOR	100,0%	100,0%	Sulfide:quinone oxidoreductase deficiency, 619221

SRD5A2	100,0%	100,0%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	100,0%	100,0%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SSR4	100,0%	100,0%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	95,8%	95,2%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98,7%	98,7%	Salt and pepper developmental regression syndrome, 609056
STAR	100,0%	100,0%	Lipoid adrenal hyperplasia, 201710
STS	97,4%	97,3%	Ichthyosis, X-linked, 308100
STT3A	100,0%	100,0%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100,0%	100,0%	?Congenital disorder of glycosylation, type Ix, 615597
STX5	100,0%	100,0%	No OMIM Disease ID
SUCLA2	100,0%	99,9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100,0%	100,0%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100,0%	100,0%	No OMIM Disease ID
SUGCT	100,0%	100,0%	Glutaric aciduria III, 231690
SUMF1	100,0%	100,0%	Multiple sulfatase deficiency, 272200
SUOX	100,0%	100,0%	Sulfite oxidase deficiency, 272300
TAZ	100,0%	100,0%	Barth syndrome, 302060
TALDO1	100,0%	100,0%	Transaldolase deficiency, 606003
TANGO2	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	100,0%	100,0%	Tyrosinemia, type II, 276600
TBXAS1	100,0%	100,0%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	100,0%	100,0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100,0%	100,0%	Transcobalamin II deficiency, 275350
TECR	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 14, 614020
TH	100,0%	100,0%	Segawa syndrome, recessive, 605407
TIMM50	100,0%	100,0%	3-methylglutaconic aciduria, type IX, 617698
TK2	100,0%	100,0%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100,0%	100,0%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98,8%	98,7%	Short stature, developmental delay, and congenital heart defects, 617044
TMEM106B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	100,0%	100,0%	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	100,0%	100,0%	Congenital disorder of glycosylation, type IIp, 616829

TMEM70	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	99,6%	99,5%	No OMIM Disease ID
TPI1	100,0%	100,0%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	100,0%	100,0%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPMT	100,0%	100,0%	No OMIM Disease ID
TPP1	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAK1	100,0%	100,0%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC2L	100,0%	100,0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TREH	100,0%	100,0%	Trehalase deficiency, 612119
TUSC3	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 7, 611093
TYMP	100,0%	100,0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	100,0%	100,0%	No OMIM Disease ID
TYR	100,0%	100,0%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100
TYRP1	100,0%	100,0%	Albinism, oculocutaneous, type III, 203290
UFM1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 14, 617899
UGT1A1	100,0%	100,0%	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
UMPS	97,0%	97,0%	Orotic aciduria, 258900
UPB1	100,0%	100,0%	Beta-ureidopropionase deficiency, 613161
UROC1	100,0%	100,0%	?Urocanase deficiency, 276880
UROD	100,0%	100,0%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100,0%	100,0%	Porphyria, congenital erythropoietic, 263700
VMA21	100,0%	100,0%	Myopathy, X-linked, with excessive autophagy, 310440
VPS13B	99,5%	99,4%	Cohen syndrome, 216550
VPS33A	89,9%	89,9%	Mucopolysaccharidosis-plus syndrome, 617303
XDH	100,0%	100,0%	Xanthinuria, type I, 278300
XYLT1	100,0%	99,7%	Desbuquois dysplasia 2, 615777
XYLT2	96,7%	96,7%	Spondyloocular syndrome, 605822
ZBTB11	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZMPSTE24	100,0%	100,0%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210

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.
TWIST is the chemistry used for 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors